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Rare disease: a national survey of paediatricians' experiences and needs

Yvonne Zurynski,[1,2] Aranzazu Gonzalez,[1] Marie Deverell,[1,2] Amy Phu,[1,2] Helen Leonard,[3] John Christodoulou,[4,5,6] Elizabeth Elliott,[1,2,7] on behalf of the APSU Impacts of Rare Diseases Study Partners

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For numbered affiliations see end of article.

Correspondence to
Dr Yvonne Zurynski, Australian Paediatric Surveillance Unit, Kids Research Institute, Westmead, NSW 2145, Australia; yvonne.zurynski@health.nsw.gov.au

ABSTRACT

Objective To describe the experiences of Australian paediatricians while caring for children with rare diseases, and their educational and resource needs.

Design A brief online survey was developed and deployed to a representative sample of 679 paediatricians from the Australian Paediatric Surveillance Unit database.

Results Of the 679 paediatricians, 242 (36%) completed the survey. The respondents were representative of all states and territories of Australia, urban and rural regions, and hospital and private practice. Almost all respondents (93%) had seen children with one or more of >350 different rare diseases during their career; 74% had seen a new patient with rare disease in the last 6 months. The most common problems encountered while caring for patients were: diagnostic delays (65%), lack of available treatments (40%), clinical guidelines (36%) and uncertainty where to refer for peer support (35%). Few paediatricians said that rare diseases were adequately covered during university (40%) or the Fellowship of the Royal Australasian College of Physicians (50%) training, and 28% felt unprepared to care for patients with rare diseases. Paediatricians wanted lists of specialist referral services (82%) and online educational modules about rare diseases (78%) that could be accessed via one online portal that consolidated multiple resources. Smartphone applications on rare diseases were favoured by paediatricians aged <50 years and by female paediatricians.

Conclusions An online educational portal should be developed and maintained for accuracy and currency of information to support dissemination of rare disease guidelines, referral pathways and coordination services relevant to Australian paediatricians and other health professionals who care for children with rare diseases.

What is already known on this topic?

- Parents of children living with rare diseases report that doctors lack knowledge about rare diseases.
- Diagnostic delays are common in children with rare diseases.
- There have been no studies of the needs of paediatricians who care for children with rare diseases.

What this paper adds?

- A study of paediatricians' knowledge, attitudes, practice and educational needs regarding rare childhood diseases.
- Paediatricians commonly see children with rare diseases but report difficulties including reaching a definitive diagnosis, lack of available treatments and guidelines, and uncertain referral pathways.
- Paediatricians called for better education about rare diseases, including a one-stop online hub where evidence-based resources could be deposited and accessed.

INTRODUCTION

Specialist paediatricians play a pivotal role in identifying, diagnosing and treating children with rare diseases.[1] Collectively, rare diseases are common, with approximately 8000 known rare diseases mostly with onset in childhood.[1–3] Children living with rare diseases often require specialist care from large teams of health professionals because of disease complexity and associated intellectual and physical disability.

Patients with rare diseases often face lengthy diagnostic delays, some families seeing between 3 and 10 different doctors and waiting >5 years for a definitive diagnosis.[4–6] We found that approximately 30% of Australian families who experienced diagnostic delays attribute delays to lack of health professionals' knowledge and difficulties accessing specialised genetic testing within a reasonable time frame.[4 5 7] Delayed diagnosis limits access to early intervention programme, could delay appropriate treatment and might result in patients receiving the wrong treatment.[5 7]

The burden of rare diseases on tertiary paediatric hospitals is significant.[4 8–10] Their complexity and chronicity means that children need ongoing multidisciplinary care from paediatric specialists, allied health professionals and disability services. Furthermore, paediatric generalists often provide

routine care, review children between visits to specialist clinics, coordinate care and organise appropriate referrals.[1 11 12] General paediatricians in rural Australia, where health services are limited, may experience problems when managing children with rare diseases, including obtaining specialist advice and accessing genetic testing and imaging technologies.[13]

To our knowledge there is only one report about the experiences of doctors while caring for patients with rare diseases.[14] That report, published by a pharmaceutical company, included a sample of 50 physicians (4 paediatricians) in the USA and 50 physicians in the UK (25 paediatricians).[14] It is unlikely to be representative given the small sample sizes and a lack of description of how physicians were selected and recruited.[14]

Our study aims to describe paediatricians' clinical practice regarding rare diseases, the difficulties they encounter, their information-seeking behaviours and their educational needs and preferences. Such information is needed to inform clinical guidelines, educational initiatives and new models of healthcare for children with rare diseases.

METHODS

Survey development

We developed a brief survey for paediatricians which asked about the following:

- gender, age, state/territory and postcode of practice, year of attainment of the Fellowship of the Royal Australasian College of Physicians (FRACP) or equivalent;
- practice setting (hospital/private practice), practice type (general or subspecialty paediatrics, remoteness (metropolitan, rural, remote);[15]
- clinical experience with rare disease patients;
- difficulties encountered when looking after children with rare diseases;
- education received about rare diseases;
- current use of information resources in clinical practice;
- preferred mode of delivery for educational resources (face-to-face, printed materials, online modules).

The survey included structured response questions with multiple tick options and Likert rating scales. Free-text answers were collected for questions about difficulties encountered when diagnosing or managing patients with rare diseases. The draft survey was reviewed for relevance by all authors and leaders of parent/peer support organisations including Genetic Alliance Australia, formerly the Association of Genetic Support of Australasia (AGSA); the Steve Waugh Foundation and SMILE Foundation (now Variety, The Children's Charity). A convenience sample of 10 paediatricians trialled the survey and provided feedback about relevance, clarity, gaps and redundancies of questions, and survey length. All feedback was incorporated into the final version (online Supplementary file) and an e-survey form was developed using the Research Electronic Data Capture Software (REDCap).[16]

Participants

In March 2015, the survey was sent to 679 paediatricians selected from the Australian Paediatric Surveillance Unit (APSU) database, which included 1366 paediatricians at that time. APSU recruits Fellows in Paediatrics and Child Health of the Royal Australasian College of Physicians (FRACP) as they graduate, whether or not they

Table 1 Characteristics of reporting clinicians and their practice

Variable		N (%)
Gender	Man	116 (48)
	Woman	126 (52)
Age groups	≤50	137 (57)
	>50	104 (43)
Year of FRACP award or equivalent	Pre 1980	9 (4)
	1980–1999	94 (39)
	≥2000	138 (57)
State of practice	New South Wales	85 (35)
	Victoria	65 (27)
	Queensland	40 (17)
	Western Australia	25 (10)
	South Australia	11 (5)
	ACT	8 (3)
	Tasmania	3 (1)
	Northern territory	2 (1)
ASGC postcode classification of remoteness	Metropolitan (RA 1)	205 (85)
	Rural (RA 2–3)	32 (13)
	Remote (RA 4–5)	2 (1)
Specialty	Paediatric generalist	130 (54)
	Subspecialist	111 (46)
Practice type	Hospital-based	135 (56)
	Private practice	19 (8)
	Hospital and private practice	85 (35)
Number of new patients seen in a typical week	<1	4 (2)
	1–5	79 (33)
	5–10	92 (38)
	10-20	46 (19)
	>20	19 (8)
Number of rare disease patients seen during clinical career	1–5	22 (10)
	6–10	35 (16)
	11–20	29 (13)
	>20	69 (31)
	>100	66 (30)

ASGC, Australian Standrad Geographical Classification; FRACP, Fellowship of the Royal Australasian College of Physicians.

Table 2 Rare disease categories seen most frequently by paediatricians during their career

Type of rare disease	N (%)	Examples of specific diseases
Metabolic/genetic	198 (88)	Galactosaemia; medium-chain acyl-CoA dehydrogenase deficiency; phenylketonuria; citrullaemia; ornithine transcarbamylase deficiency; trisomies 8, 13, 18
Neurological	161 (72)	Lissencephaly; Lennox-Gastaut syndrome; muscular dystrophies; neuraxonal dystrophy
Immunological	102 (45)	Severe combined immunodeficiency; IgA deficiency; hyper IgE syndrome; chronic granulomatous disease
Skeletal	95 (42)	Achondroplasia; osteogenesis imperfecta; osteopetrosis; asphyxiating thoracic dystrophy
Cardiac	85 (38)	Ebstein's anomaly; hypoplastic left heart syndrome; transposition of the great arteries; long QT syndrome; cardiomyopathy
Respiratory	80 (36)	Alveolar capillary dysplasia; pulmonary lymphangiectasis; surfactant protein C deficiency
Endocrine	80 (36)	Congenital hypothyroidism; McCune-Albright syndrome; hypophosphatasia; congenital adrenal hyperplasia
Gastrointestinal	77 (34)	Biliary atresia; giant omphalocoele; gut duplication; Neonatal haemochromatosis; Crohn's disease
Renal	75 (33)	Polycystic kidneys; congenital nephrotic syndrome; nephronophthisis; Bartter syndrome, renal tubular acidosis
Dermatological	75 (33)	Epidermolysis bullosa; incontinentia pigmenti; Stevens-Johnson syndrome; toxic epidermal necrolysis
Connective tissue	73 (32)	Pseudoxanthoma elasticum; Marfan syndrome; Ehlers-Danlos syndrome
Cancer	71 (32)	Medulloblastoma; Wilms; retinoblastoma; atypical teratoid rhabdoid tumour; Ewing's sarcoma; hepatoblastoma; childhood melanoma
Infectious	52 (23)	Kingella kingae infection; HIV infection; onchocerciasis; respiratory papillomatosis
Rheumatic	39 (17)	Juvenile onset rheumatoid arthritis; systemic lupus erythematosus; Behçet's disease

CoA, coenzyme A.

have an interest in rare diseases. About 54% are general paediatricians, and the rest are paediatric subspecialists (emergency physicians, endocrinologists, neonatologists, geneticists and so on) paediatric surgeons and child psychiatrists.[17 18]

In this paper we refer to all respondents as paediatricians. To obtain a representative sample, the APSU list was stratified by state/territory jurisdiction, ordered alphabetically within each state/territory and every second paediatrician was selected to participate. The 10 paediatricians who piloted the survey were excluded.

Procedure

An email containing a personalised web link to the survey was sent. No identifying information was collected apart from the paediatrician's APSU ID number, which was used to track responses. Reminder emails were sent to non-responders 2 and 4 weeks after the initial email.

Data analysis

Data were exported from REDCap into the Statistical Package for Social Science (IBM SPSS) and analysed using descriptive statistics: mean, SD, frequencies and percentages. For non-parametric variables, median and IQR was calculated. Associations between groups were tested using the X^2 test.

Ethics approval

Ethics approval and research governance approval were obtained from the Sydney Children's Hospitals Network Human Research Ethics Committee (LNR/14/SCHN/496; LNRSSA/14/SCHN/505).

RESULTS

Demographics and clinical experience with rare diseases

Of paediatricians surveyed 242 (36%), completed the survey, with representation from all states/territories (table 1). There were 52% women, 57% were <51 years old, 57% received their FRACP in 2000 or later and over a half (54%) were general paediatricians (table 1). The majority (85%) practised in a metropolitan area, and 56% in a hospital setting (table 1). There was no significant difference between responders and non-responders on state/territory or specialty. The demographic details of the responders match with previously published APSU data.[18] Almost all (98%) had reliable internet access. Interestingly, the three with unreliable internet access were based in a metropolitan centre. There was little difference in the use of smartphones and computer tablets in everyday clinical practice between paediatricians based in rural or remote regions (79%) and metropolitan settings (74%).

Table 3 Difficulties encountered by paediatricians while caring for children with rare diseases

Areas of difficulty	n=238(%)
Delay in, or inability to make a definitive diagnosis	155 (65)
Lack of available treatments	94 (40)
Lack of treatment/management guidelines	86 (36)
Lack of diagnostic guidelines	83 (35)
Uncertainty about available peer support groups for the patient and family	82 (35)
Lack of access to diagnostic tests	73 (31)
Difficulties in accessing specialised clinics/services	68 (29)
Difficulties in accessing genetic testing/services	65 (27)
Difficulties in accessing allied health services	62 (26)
Uncertainty about where to refer to	49 (21)
Difficulty accessing new drugs or therapies currently available overseas, not yet licensed in Australia	46 (19)
Other difficulties	14 (6)

Most (89%) of respondents believed that ≤5% of the child population has a rare disease. Most (71%) respondents saw 1–10, 19% saw 10–20, and 8% saw >20 new patients per week (table 1). Almost all respondents (93%) had seen at least one child with a rare disease during their clinical career. About two-thirds (60%) had seen >20 such children during their career (table 1) and most of these were paediatric subspecialists including geneticists, neonatologists, neurologists or surgeons. Over 350 different rare diseases were seen (table 2). Paediatricians who had seen >100 rare disease patients in their clinical career were more likely to have completed their FRACP before 2000 (χ^2= 4.66; p=0.03). The majority of respondents (97%) reported caring for children with undiagnosed clusters of signs and symptoms, with 57% seeing ≥10 such patients during their clinical career. In total, 238 (98%) clinicians had seen children with rare diseases and/or undiagnosed clusters of signs and symptoms, and of these, 97% had seen a new case in the last 3 years and 74% had seen a new case in the last 6 months.

Almost all paediatricians (98%) reported encountering at least one difficulty when caring for children with rare diseases, most commonly delays in reaching a definitive diagnosis, lack of available treatments, lack of clinical guidelines, uncertainty about existence of peer support groups for patients and parents and lack of access to diagnostic tests (table 3). Other difficulties included accessing allied health services, uncertainties about referral pathways, accessing genetic testing and accessing drugs currently available overseas, but not licensed in Australia (table 3).

Confidence about ability to care for children with rare diseases

Almost all (99%) paediatricians acknowledged rare diseases are important, 72% felt prepared to look after these patients, and 62% were confident about where to find information about rare diseases. Most paediatricians (92%) said that they would use a directory of specialist services if one was available, and 64% were confident about referral pathways. The majority (87%) believed that patients with rare disease are best looked after in a multidisciplinary clinic and 79% said they had adequate access to rare disease experts.

Awareness and use of educational resources

Less than half (40%) of paediatricians said rare diseases were adequately covered in their medical degree, while 50% said they were adequately covered during FRACP training. Paediatricians reported using the following sources of information in everyday clinical practice: consultation with colleagues (92%); web-based resources (91%); textbooks (49%) and mobile phone or tablet applications (apps) (30%).

All respondents were aware of Medline (100%), and almost all were aware of the Cochrane Library, Up-To-Date and National Institutes for Healthcare Excellence (NICE) guidelines (table 4). Few respondents were aware of rare disease resources. The best known included APSU protocols, case definitions and website, Online Mendelian Inheritance in Man, Pictures Of Standard Syndromes and Undiagnosed Malformations and the portal for rare diseases and orphan drugs (Orphanet) (table 4). Just over 40% of respondents had attended lectures or workshops on rare diseases in the last 5 years, and most found these useful in terms of content (93%), resources (61%) and networking opportunities (59%) (table 4).

Paediatricians' needs for education and resources to support practice

Respondents favoured web portals providing fact sheets, lists of peer support groups for patients and lists of specialist services for referral (table 5). Three-quarters indicated they would use educational modules about rare diseases if offered via the APSU or RACP websites. Smartphone/tablet apps supporting rare disease diagnosis and management were favoured by 72% (table 5). Paediatricians aged ≤50 years were significantly more likely to access smartphone/tablet apps on rare disease diagnosis and clinical management (X^2=13.5; p<0.001), as were female paediatricians (X^2=6.9; p<0.01).

Factors that would increase the use of information resources included up to date clinically relevant content (97%), access through a single web portal (93%) and free or low cost (93%).

DISCUSSION

This is the first study of paediatricians' experiences of providing care to patients with rare diseases using a

Table 4 Awareness and usefulness of rare disease internet resources

Internet resource	N*	Aware of resource N (%)	Aware and used resource N (%)	Aware, used and found resource useful N (%)
Rare disease-specific resources				
APSU Study protocols, case definitions, website and report	239	213 (90)	128 (54)	50 (21)
OMIM	234	178 (76)	118 (50)	73 (31)
POSSUM	233	157(67)	64 (28)	30 (13)
Orphanet Portal for Rare Diseases	234	116 (50)	66 (28)	45 (19)
Orphanet Journal of Rare Diseases	231	92 (40)	43 (19)	26 (11)
HGSA	226	89 (39)	25 (11)	9 (4)
NORD, USA	235	83 (35)	26 (11)	13 (6)
AGSA†	225	50 (22)	15 (7)	7 (3)
Centre for Genetic Education (NSW Health)	227	49 (22)	28 (12)	20 (9)
EURORDIS	228	47 (21)	15 (7)	5 (2)
General resources				
PubMed, Medline, or similar	240	240 (100)	235 (98)	168 (71)
Cochrane Library	234	233 (100)	208 (89)	127 (61)
Up-To-Date	239	233 (98)	203 (85)	147 (72)
BMJ Best Practice guidelines	237	216 (91)	136 (57)	62 (46)
NICE, UK	236	222 (94)	182 (77)	117 (64)
RACP website and CME Resources	231	199 (87)	107 (46)	23 (22)
Centres for Disease Control National Guidelines Clearing House	235	120 (51)	49 (21)	18 (37)

*Number of respondents that answered this question.
†AGSA changed its name to Genetic Alliance Australia.
AGSA, Association of Genetic Support of Australasia; EURORDIS; CME, continuing medical education; European Organisation for Rare Diseases; HGSA, Human Genetics Society of Australia; NICE, National Institutes for Health and Clinical Excellence; NORD, National Organisation for Rare Diseases; OMIM, Online Mendelian Inheritance in Man; POSSUM, Pictures Of Standard Syndromes and Undiagnosed Malformations; RACP, Royal Australasian College of Physicians.

representative, systematically recruited sample (n=242). Almost all (98%) of paediatricians in our survey had encountered patients with rare diseases or unusual undiagnosed syndromes in their clinical career. Approximately one-third had seen ≥100 diagnosed cases representing a wide variety of conditions. Almost three-quarters saw a new case in the last 6 months. Our results align with the reported high economic burden of genetic disorders in a population-based study in Western Australia.[8]

Paediatricians experienced problems when caring for children with rare diseases, most commonly delay or inability to make a definitive diagnosis. This was attributed to difficulties accessing diagnostic tests including genetic tests. In Australia, the number of tests covered by the publicly funded Medicare Benefits Scheme has remained static but new tests have become available, many of which are not covered, leading to potential inequitable access to genetic testing according to the individual's financial situation.[19] Similar concerns about access to genetic tests have been reported by patients' families.[5 6] Paediatricians also reported delays in interpretation of genetic tests, suggesting a need for increased clinical genetics capacity in Australia. The burden on clinical geneticists will undoubtedly increase with availability of cheaper sequencing, however delays in clinical interpretation of variants and diagnostic delays should ultimately diminish.[20]

The lack of clinical guidelines, lack of treatment options and inability to access drugs that there are available overseas but not licensed in Australia also frustrates paediatricians. Paediatricians reported difficulties in accessing allied health services and uncertainties about referral pathways. Parents also report problems in accessing treatments, allied health services and care coordination.[4–6]

A report by the pharmaceutical company Shire involving 50 physicians from the USA and 50 from UK showed that they needed to see rare disease patients more frequently than other patients for diagnosis and monitoring and found coordinating their care difficult.[14] This concurs with our studies of paediatricians and parents.[4–6] A large proportion of physicians in the Shire report (USA 86%; UK 90%) reported difficulties accessing treatments, compared with only 40% in our study. This stark difference is not easily explained given that more rare drugs

Table 5 Likelihood of paediatricians using different types of educational resources, if they were available

Type of resource	N*	Likely to use N (%)	Unlikely to use N (%)
A web portal providing fact sheets about specific rare diseases which you could give to your patients	239	228 (95)	11 (5)
A web portal providing a listing of support groups available for your patients and their families	238	222 (93)	16 (7)
A web portal providing a listing of specialists and specialist clinics you can refer your patients to	238	196 (82)	42 (18)
Online modules via the APSU or RACP about specific rare diseases or groups of rare diseases	240	188 (78)	52 (22)
Online modules via the APSU or RACP about how to use already existing online resources	237	166 (70)	71 (30)
Smartphone/tablet applications on rare disease diagnosis and management	232	166 (72)	66 (28)
Printed materials/modules about specific rare diseases or groups of rare diseases	234	160 (68)	74 (32)
Smartphone/tablet applications about how to use already existing online resources	233	153 (66)	80 (34)
Face-to-face educational workshops/seminars about specific rare diseases or groups of rare diseases	235	149 (63)	86 (37)
Face-to-face educational workshops/seminars about how to use already existing on-line resources	237	122 (51)	115 (49)

*Number of respondents that answered this question.
APSU, Australian Paediatric Surveillance Unit; RACP, Royal Australasian College of Physicians.

are licensed in the USA and UK than in Australia. The Shire report had a small sample and did not describe the selection, recruitment and representativeness of the respondents, making comparisons difficult.[14]

Our study is the first to highlight paediatricians' awareness and use of educational resources, specifically when caring for patients with rare diseases. Only 57% said their medical degree prepared them to recognise patients with rare diseases and one-third felt 'unprepared' to care for children with rare diseases. The French National Plan for Rare Diseases stipulates that the undergraduate medical curriculum must include specific education in rare diseases, including how to access reliable information.[21] There is no such requirement in Australia, and almost 40% of respondents in our study lacked confidence about finding reliable information about rare diseases.

The majority were aware of, and used online educational resources including Medline, the Cochrane Library, and the NICE guidelines to support their clinical practice. Fewer were aware of, or had used, rare disease specific resources such as OMIM, POSSUM and the Orphanet portal. Only 35% were aware of the NORD portal, which houses 'Physician Guides' for over 1200 rare diseases[22] and only 22% were aware of the New South Wales Health Centre for Genetics Education.[23]

Almost 90% were aware of APSU resources on rare diseases for example, case definitions, surveillance protocols and reports and just over half had used these resources. This high level of awareness of APSU resources was likely biased as the sample was drawn from the APSU. The relatively low use of APSU resources may depend on the paediatrician seeing a child with one of the 60 rare conditions for which APSU provides resources. APSU plans to extend the reach and usefulness of these resources for clinicians.

The APSU database was estimated to represent ~90% of Australian paediatricians in active clinical practice.[17 18] The relatively small response fraction and the likelihood of response from paediatricians who are interested in, or have exposure to patients with rare diseases, might have introduced some bias. Nevertheless the sample was large and representative across states/territories, specialties and subspecialties, and the response fraction is similar to other surveys of busy health professionals.

Paediatricians called for rare disease resources to be made available via a single portal to simplify searching for information. The majority (82%) wanted a directory of specialist referral services for rare diseases, despite only 21% being uncertain about referral pathways. Importantly, over 90% of paediatricians called for easily available printable fact sheets for patients and families, and a directory of family support groups to which families could be referred. Although Genetic Alliance Australia (formerly AGSA) provides counselling services and links to peer support groups for many genetic diseases, few paediatricians knew about this valuable service.

Paediatricians said they would be more likely to use online resources than printed materials or face-to-face opportunities for education. Availability of online resources and awareness of these is essential to support

their use in clinical practice, as needed. Female paediatricians and paediatricians aged <50 years would be more likely to use smartphone apps in clinical practice and these groups should be targeted if rare diseases apps were to be developed.

The APSU website houses information about many rare diseases, and provides links to well-respected resources such as OMIM and the Orphanet portal. Because APSU is well known to many Australian paediatricians and other child health clinicians, there is opportunity to build on existing infrastructure to develop a comprehensive information and educational portal with links to high-quality resources nationally and internationally. Furthermore, there is an opportunity to pool resources and to reach paediatricians in at least 11 countries through the International Network of Paediatric Surveillance Units.[24]

Our results support the need to raise awareness of rare disease resources while consolidating and disseminating via a single portal. To improve awareness, access to and actual use of resources in clinical practice, multiple strategies are needed to engage with paediatricians. Embedding specific teaching about rare diseases in undergraduate and postgraduate medical curricula would better equip future paediatricians to care for children with rare diseases. Paediatricians also need access to affordable genetic tests for their patients and to clinical geneticists' expertise to support meaningful clinical decisions.

Author affiliations
[1]Australian Paediatric Surveillance Unit, Kids Research Institute, Westmead, New South Wales, Australia
[2]Discipline of Child and Adolescent Health, Sydney Medical School, The University of Sydney, Sydney, New South Wales, Australia
[3]Telethon Kids Institute, The University of Western Australia, West Perth, Western Australia, Australia
[4]Murdoch Children's Research Institute, University of Melbourne, Melbourne, Victoria, Australia
[5]Clinical School, Sydney Children's Hospital Network, Sydney, New South Wales, Australia
[6]Genetic Metabolic Disorders Research Unit, Western Sydney Genetics Program, the Children's Hospital at Westmead, Westmead, New South Wales, Australia
[7]Department of Paediatrics, Faculty of Medicine, Dentistry and Health Sciences, University of Melbourne, Victoria, Australia

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Contributors YZ: designed the study and analysis, interpreted data, and drafted the manuscript. EE, JC, HL and MD: contributed to study design and edited the manuscript. AG: finalised the survey design, undertook data analysis and wrote the results section. AP: digitised the survey, randomised the sample of respondents, oversaw the data collection, cleaned the data and prepared it for analysis. All authors: read and approved the final manuscript.

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REFERENCES

1. Zurynski Y, Frith K, Leonard H, *et al*. Rare childhood diseases: how should we respond? *Arch Dis Child* 2008;93:1071–4.
2. Elliott E, Zurynski Y. Rare diseases are a 'common' problem for clinicians. *Aust Fam Physician* 2015;44:630–3.
3. EURORDIS. *The voice of 12,000 patients: experiences and expectations of rare disease patients on diagnosis and care in Europe*. Boulogne-Billancourt, France: Eurordis, 2009:1–324.
4. Anderson M, Elliott EJ, Zurynski YA. Australian families living with rare disease: experiences of diagnosis, health services use and needs for psychosocial support. *Orphanet J Rare Dis* 2013;8:22.
5. Zurynski Y, Deverell M, Dalkeith T, *et al*. Australian children living with rare diseases: experiences of diagnosis and perceived consequences of diagnostic delays. *Orphanet J Rare Dis* 2017;12:68.
6. Pelentsov LJ, Fielder AL, Laws TA, *et al*. The supportive care needs of parents with a child with a rare disease: results of an online survey. *BMC Fam Pract* 2016;17:88.
7. Baynam G, Pachter N, McKenzie F, *et al*. The rare and undiagnosed diseases diagnostic service - application of massively parallel sequencing in a state-wide clinical service. *Orphanet J Rare Dis* 2016;11:77.
8. Walker CE, Mahede T, Davis G, *et al*. The collective impact of rare diseases in Western Australia: an estimate using a population-based cohort. *Genet Med* 2017;19:546–52.
9. Zurynski YA, Peadon E, Bower C, *et al*. Impacts of national surveillance for uncommon conditions in childhood. *J Paediatr Child Health* 2007;43:724–31.
10. McCandless SE, Brunger JW, Cassidy SB. The burden of genetic disease on inpatient care in a children's hospital. *Am J Hum Genet* 2004;74:121–7.
11. Knight AW, Senior TP. The common problem of rare disease in general practice. *Med J Aust* 2006;185:82–3.
12. Phillips WR. Zebras on the commons: rare conditions in family practice. *J Am Board Fam Pract* 2004;17:283–6.
13. Australian Government Department of Health. National strategic framework for rural and remote health. 2011 http://www.health.gov.au/internet/main/publishing.nsf/content/national-strategic-framework-rural-remote-health (accessed 9 Jun 2017).
14. Shire. Rare disease impact report: insights from patients and the medical community. 2013 https://globalgenes.org/wp-content/uploads/2013/04/ShireReport-1.pdf (accessed 9 June 2017).
15. Australian Bureau of Statistics. Australian standard geographical classification. 2011 http://www.abs.gov.au/websitedbs/D3310114.nsf/home/Australian+Standard+Geographical+Classification+%28ASGC%29 (accessed 9 June 2017).
16. Harris PA, Taylor R, Thielke R, *et al*. Research electronic data capture (REDCap)-a metadata-driven methodology and workflow process for providing translational research informatics support. *J Biomed Inform* 2009;42:377–81.
17. He S, Zurynski YA, Elliott EJ. Evaluation of a national resource to identify and study rare diseases: the Australian Paediatric Surveillance Unit. *J Paediatr Child Health* 2009;45:498–504.

18. He S, Zurynski YA, Elliott EJ. What do paediatricians think of the Australian Paediatric Surveillance Unit? *J Paediatr Child Health* 2010;46:412–8.
19. Mina K, Suthers G. The spectrum of genetic testing in Australia. *Pathology* 2013;45(Suppl 1):S3.
20. Beale S, Sanderson D, Sanniti A, *et al*. A scoping study to explore the cost-effectiveness of next-generation sequencing compared with traditional genetic testing for the diagnosis of learning disabilities in children. *Health Technol Assess* 2015;19:1–90.
21. Ayme S, Rodwell C, eds. *2013 Report on the state of the art of rare disease activities in Europe*. Brussels: European Union, 2013. http://www.eucerd.eu/upload/file/Reports/2013ReportStateofArtRDActivities.pdf. (assessed 9 Jun 2017).
22. National Organization for Rare Disorders. NORD Physician Guides: rare disease resources for medical professionals. http://nordphysicianguides.org/index-of-guides (assessed 9 Jun 2017).
23. NSW Government Health. Centre for genetics education. Resources for health professionals. http://www.genetics.edu.au/Professionals (assessed 9 Jun 2017).
24. Zurynski Y, Grenier D, Lynne R, eds. International network of paediatric surveillance units: 15 years of international research into rare childhood diseases. INoPSU, 2013. http://www.inopsu.com/Publications

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