Paediatricians Caring for Children with Rare Diseases: Survey of knowledge, practice and future educational needs

Thank you for agreeing to complete this survey which will take 5-15mins to complete.
The survey is anonymous.

Please click on the most appropriate answer to each question. If you wish to change your choice please click on "reset" on the far right of each question.
For some questions you will be asked to comment - please write your comments in the box provided.
At the end of the survey please click "Submit".
You may also choose to save your answers and return to the survey at a later time.

By "rare disease" we mean a disease or condition which occurs infrequently (< 1in 2,000 children). Such diseases/conditions often include, but are not limited to, rare genetic syndromes, rare infectious diseases, or they may include very rare complications of more common diseases e.g. rhabdomyolysis due to influenza.

About you and your practice

1. Gender:

☐ Male ☐ Female

2. Age:

☐ < 30 ☐ 30-35 ☐ 36-40 ☐ 41-45 ☐ 46-50 ☐ 51-55 ☐ 56-60 ☐ 61-64 ☐ >65

3. In which year were you awarded your FRACP (Paediatrics and Child Health) or equivalent?

_____

4. In which State or Territory do you currently practice?

☐ NSW ☐ VIC ☐ QLD ☐ WA ☐ SA ☐ NT ☐ TAS ☐ ACT

5. Is your practice:

☐ hospital based only ☐ private practice only ☐ combination of hospital and private practice

6. What is the postcode of your main place of practice? _____

7.What category best describes your current practice?

☐ General Paediatrics only
☐ General Paediatrics with one or more specialties (e.g. Developmental, Respiratory)
☐ Subspecialty Paediatrics only (no general Paediatrics)
☐ Paediatric Surgery
☐ Other

Please specify specialty: _____

Please specify specialty: _____

Please specify: _____

8. Please estimate how many NEW patients you see in your practice in an average week

☐ < 1 / week ☐ 1-5 / week ☐ 5-10 /week ☐ 10-20 /week ☐ >20 / week

Your Experience with Rare Disease Patients

9. What proportion of the general Australian child population do you believe is affected by rare diseases?

☐ < 1% ☐ 2-3% ☐ 3-5% ☐ 6-8% ☐ 9-10% ☐ >10%

10. Have you ever looked after a patient(s) with a diagnosed rare disease or condition?

☐ Yes ☐ No ☐ Not sure

11. To the best of your knowledge, during your clinical career as a Consultant, please estimate how many of your patients have had a diagnosed rare disease/condition?

☐ 1-5 ☐ 5-10 ☐ 10-20 ☐ >20 ☐ >100

12. To the best of your recall, what types of rare conditions have you been involved in managing? (tick as many as apply)

☐ Metabolic/Genetic
☐ Renal
☐ Gastrointestinal
☐ Endocrine
☐ Neurological
☐ Connective tissue
☐ Rheumatic
☐ Skeletal
☐ Respiratory
☐ Infectious
☐ Immunological
☐ Dermatological
☐ Cardiac
☐ Cancer

Please provide examples of diseases you have looked after

13. Have you ever looked after a patient(s) with an unusual cluster of signs and symptoms for whom a definitive diagnosis was hard to establish?

☐ Yes ☐ No ☐ Not Sure

14. To the best of your recollection, can you estimate how many such patients who have no definitive diagnosis you have managed throughout your clinical career as a Consultant?

☐ 1-5 ☐ 5-10 ☐ 10-20 ☐ >20

15. When did you last see a new patient with EITHER a diagnosed rare disease or an undiagnosed cluster of signs and symptoms?

☐ < 6 months ago ☐ 6-12 months ago ☐ 1-3 years ago ☐ >3 years ago

16. When diagnosing and/or managing patients with rare diseases or conditions, in which of the following areas have you encountered difficulties? (select as many as apply)

☐ Lack of diagnostic guidelines

Comment:

☐ Lack of access to diagnostic tests

Comment:

☐ Delay in, or inability to make a definitive diagnosis

Comment:

☐ Lack of treatment/management guidelines

Comment:

☐ Lack of available treatments

Comment:

☐ Difficulty accessing new drugs or therapies currently used overseas, not yet licensed in Australia

Comment:

☐ Uncertainty about where to refer to

Comment:

☐ Difficulties in accessing specialised clinics/services

Comment:

☐ Difficulties in accessing allied health services (e.g. physio, speech, OT, psychology etc.)

Comment:

☐ Difficulties accessing genetic testing/services

Comment:

☐ Uncertainty about available peer support groups for the patient and his/her family

Comment:

☐ Other difficulties

Comment:

Your Education about Rare Diseases

17. Were rare childhood diseases covered in your original Medical Degree curriculum?

☐ Yes ☐ No ☐ Can't remember

Please comment:

18. To the best of your recollection, did your Fellowship training adequately cover rare childhood diseases, to enable you to effectively provide patient care?

☐ Yes ☐ No ☐ Can't remember

Please comment:

19. Have you attended any educational lectures/workshops about rare diseases in the last 5 years?

☐ Yes ☐ No ☐ Can't remember

Was/were these workshops/conferences useful?

☐ Yes ☐ No ☐ Can't remember

What you find most useful? (tick all that apply)

☐ Content information
☐ Networking opportunities
☐ Resources
☐ Other

Please specify

Use of Information Resources in Your Practice

20. In your everyday Paediatric practice, do you have reliable access to the internet?

☐ Yes ☐ No

21. Have you used internet resources to guide your patient management?

☐ Yes ☐ No

22. In your everyday Paediatric practice do you use a smartphone (e.g. iPhone or Blackberry) or tablet/iPad?

☐ Yes ☐ No

23. Please indicate which of the following resources you have heard of, used and found helpful in your paediatric practice? (tick across all applicable columns).

If you tick I don't know of this resource please do not tick any other column for that resource

	I don't know of this resource	I have heard of this resource	I have used this resource	I found this resource helpful
Australian Paediatric Surveillance Unit (APSU) Study protocols, case definitions, website and reports	☐	☐	☐	☐
PubMed, Medline, or similar	☐	☐	☐	☐
BMJ Best Practice guidelines	☐	☐	☐	☐
CDC National Guidelines Clearing House	☐	☐	☐	☐
National Institutes for Health and Clinical Excellence (NICE; UK)	☐	☐	☐	☐
ORPHANET portal for rare diseases	☐	☐	☐	☐
Online Mendelian Inheritance in Man (OMIM)	☐	☐	☐	☐
NORD (National Organisation for Rare Diseases)	☐	☐	☐	☐
Up-To-Date	☐	☐	☐	☐
Cochrane Library	☐	☐	☐	☐
European Organisation for Rare Diseases (EURORDIS)	☐	☐	☐	☐
POSSUM (Pictures Of Standard Syndromes and Undiagnosed Malformations)	☐	☐	☐	☐
Centre for Genetic Education (NSW Health)	☐	☐	☐	☐
Human Genetics Society of Australia (HGSA)	☐	☐	☐	☐
Association of Genetic Support of Australasia (AGSA)	☐	☐	☐	☐
RACP website and CME resources	☐	☐	☐	☐
Orphanet Journal of Rare Diseases	☐	☐	☐	☐

24. Which of the following resources do you routinely use in your everyday practice to help you diagnose and manage your paediatric patients? (select all applicable)

☐ Textbooks
☐ Consultation with colleagues
☐ Referral to specialist clinics/tertiary centres
☐ Web-based resources
☐ Apps on smartphones or other mobile devices (e.g. IPhone; Blackberry; tablet/iPad)
☐ Other

Please specify other web-based resources

Please specify other sources

25. What types of educational resources about rare diseases are you likely to use if these were available?

	Very likely	Likely	Unlikely	Very unlikely
Online modules via the APSU or RACP about how to use already existing on-line resources	☐	☐	☐	☐
Face-to-face educational workshops/seminars about how to use already existing on-line resources	☐	☐	☐	☐
Smart phone/tablet applications about how to use already existing on-line resources	☐	☐	☐	☐
Online modules via the APSU or RACP about specific rare diseases or groups of rare diseases	☐	☐	☐	☐
Face to face educational workshops/seminars about specific rare diseases or groups of rare diseases	☐	☐	☐	☐
Printed materials/modules about specific rare diseases or groups of rare diseases	☐	☐	☐	☐
Smart phone/tablet applications on rare disease diagnosis and management	☐	☐	☐	☐
A web portal providing a listing of specialists and specialist clinics you can refer your patients to	☐	☐	☐	☐
A web portal providing a listing of support groups available for your patients and their families	☐	☐	☐	☐
A web portal providing fact sheets about specific rare diseases which you could give to your patients	☐	☐	☐	☐

25. What would make you more likely to regularly access already existing web-based resources on rare diseases in your clinical practice?

	Yes	No
Know-how on what each web-based resource offers, how to access and how to use them, to optimise your clinical practice	☐	☐
A single web portal listing all available web-based resources with links to these resources	☐	☐
Fast point-of-care (mobile) access to each site (eg. iPhone application)	☐	☐
Up-to-date, clinically reliable content	☐	☐

Free access/affordable subscription	☐	☐
Regular updates on web-based resources delivered to your email	☐	☐

Your Attitudes to Rare Diseases and their Management

27. To what extent do you agree with the following statements:

	Strongly agree	Agree	Disagree	Strongly disagree
I feel that my medical degree training was adequate for me to recognise patients with rare diseases	☐	☐	☐	☐
I don't think I need training about how to use databases and other on-line resources about rare diseases	☐	☐	☐	☐
I feel confident that I would know the referral pathway when faced with a rare disease patient	☐	☐	☐	☐
I feel confident that I would know where to find information about diagnosis and management of rare diseases	☐	☐	☐	☐
Rare diseases are not important	☐	☐	☐	☐
I believe that patients with rare diseases are best looked after in a multidisciplinary clinic	☐	☐	☐	☐
I feel unprepared to look after patients with rare diseases	☐	☐	☐	☐
I have adequate access to experts in rare diseases who advise me about management of rare disease patients	☐	☐	☐	☐
I would use a directory of specialised rare disease services or clinics if such a directory was available	☐	☐	☐	☐

Thank you for taking part in this survey