
Celebrating 25 Years of the Australian Paediatric Surveillance Unit, 1993-2018
The Contribution of Paediatricians to Child Health
Elizabeth Elliott
CORPORATE HOORE

Rare diseases are common, but neglected

- 10,000 rare diseases
- ~ 400,000 children <15y

- Lack of national data, training, services, diagnostic tests, research funding, drugs, family support

We need data to advocate, inform

National Data are important

National Institutes of Health, USA
Office for Rare Diseases

"Because of the small number of affected patients in any one location, rare disease research requires the collaboration of scientists from multiple disciplines and the capacity to share access to geographically distributed, national research resources and patient populations."

- bypass state/territory health systems
- address gaps in surveillance systems

APSU HISTORY

It all began in the UK with the BPSU….1986

MD graduation….1992

Visit to Australia by Susan Hall….1993

APSU Steering Committee 1993-4

- Peter Procopis*
- Margaret Burgess
- Alan Carmichael
- Kerry Chant
- Elizabeth Elliott
- Michael Frommer
- Susan Hall
- Robert Hall
- David Isaacs
- Craig Mellis
- Elisabeth Murphy
- Terry Nolan
- Kim Oates
- John Pearne
- Don Roberton
- Fiona Stanley
- Barry Taylor
- Graeme Vimpani
- Alan Walker
- John Ziegler

* **Chair, President Australian College Paediatrics**

Clive and Vera Ramaciotti Foundation

"The first year of surveillance"

Clive and Vera Ramaciotti, Financial Markets Foundation for Children, NHMRC, ARC, Australian Government Department of Health, State Health Departs, Creswick Foundation, University Sydney Medical Foundation, Sax Institute, SMILE Foundation, Steve Waugh Foundation, Royal North Shore Hospital, Rett Association, Genzyme, CSL, Individual studies. In Kind: Children Hospital Westmead, RACP

First student, first publication, 1993

Med J Aust. 1993 Sep 6;159(5):354.

Rare disease surveillance in children.

Elliott EJ, Chant K.

PMID: 8361436 [PubMed - indexed for MEDLINE]

J Paediatr Child Health. 1994 Dec;30(6):463-5.

Rare disease surveillance.

Elliott EJ, Chant KG.

Department of Paediatrics and Child Health, Unive

DISEASE SURVEILLENCE

May 1993

The Australian Paediatric Surveillance Unit

‘National resource’ or platform for surveillance of rare chronic and complex diseases:

Rare infectious, vaccine preventable, genetic, mental health, injuries, toxins, adverse effects etc

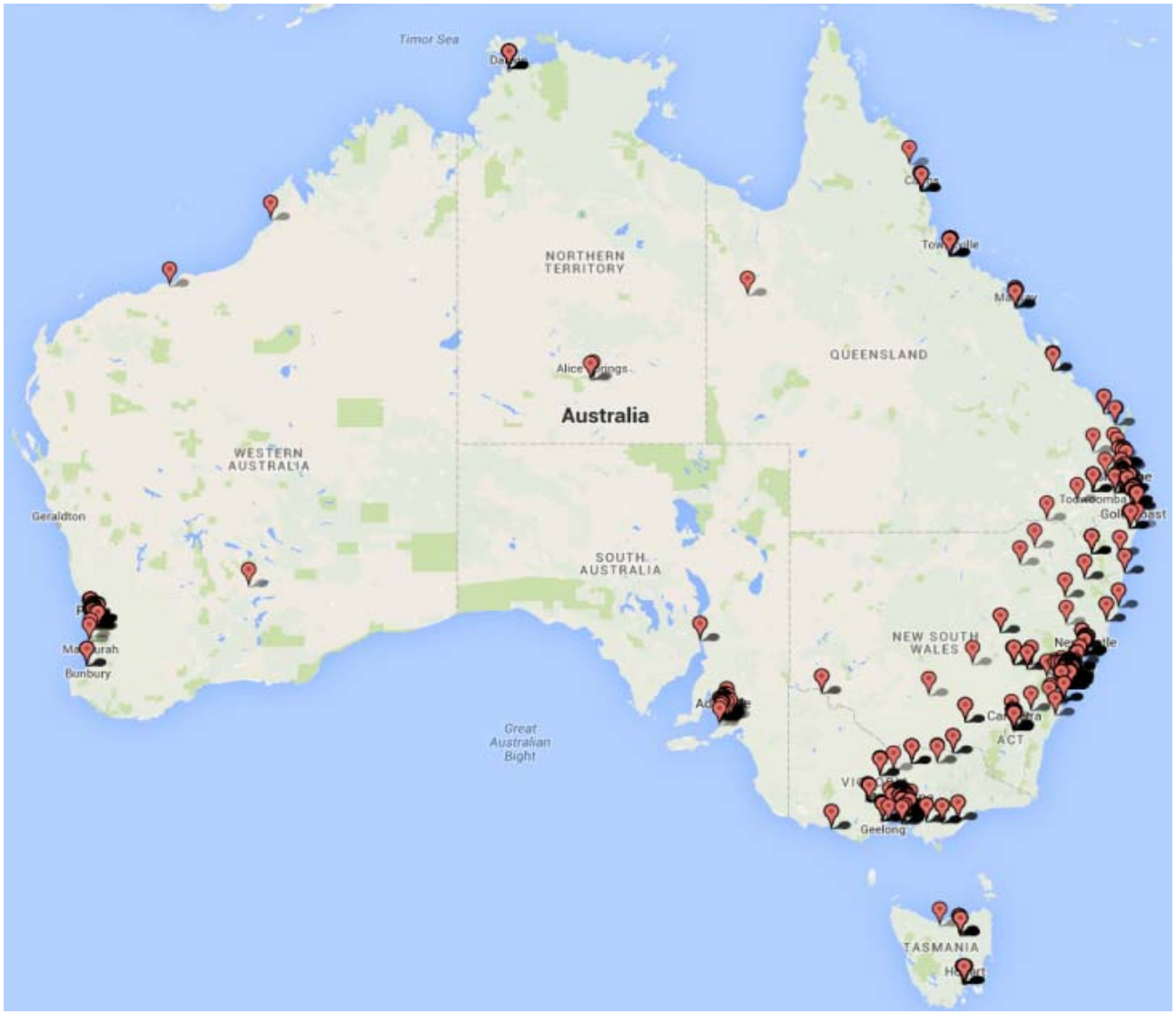
Current studies

- 22Q11.2 Deletion Syndrome
- Acute Flaccid Paralysis
- Congenital CMV infection
- Congenital Rubella syndrome
- Congenital Varicella syndrome
- Early onset eating disorder
- Fetal Alcohol Spectrum Disorder
- HIV, AIDS, Perinatal exposure
- Juvenille onset recurrent respiratory papillomatosis
- MECP2 duplication syndrome
- Microcephaly children <12m
- Neonatal varicella infection
- HSV infection in neonates, young children
- Rett syndrome
- Severe complications Influenza (June-September 2018)
- Severe injury from Disc Battery
- Stroke in children <2 years
- Vitamin K deficiency bleeding

Involves paediatricians in research

APSU – Who reports?

- All practising RACP Fellows Paediatrics & Child Health invited
- National representation

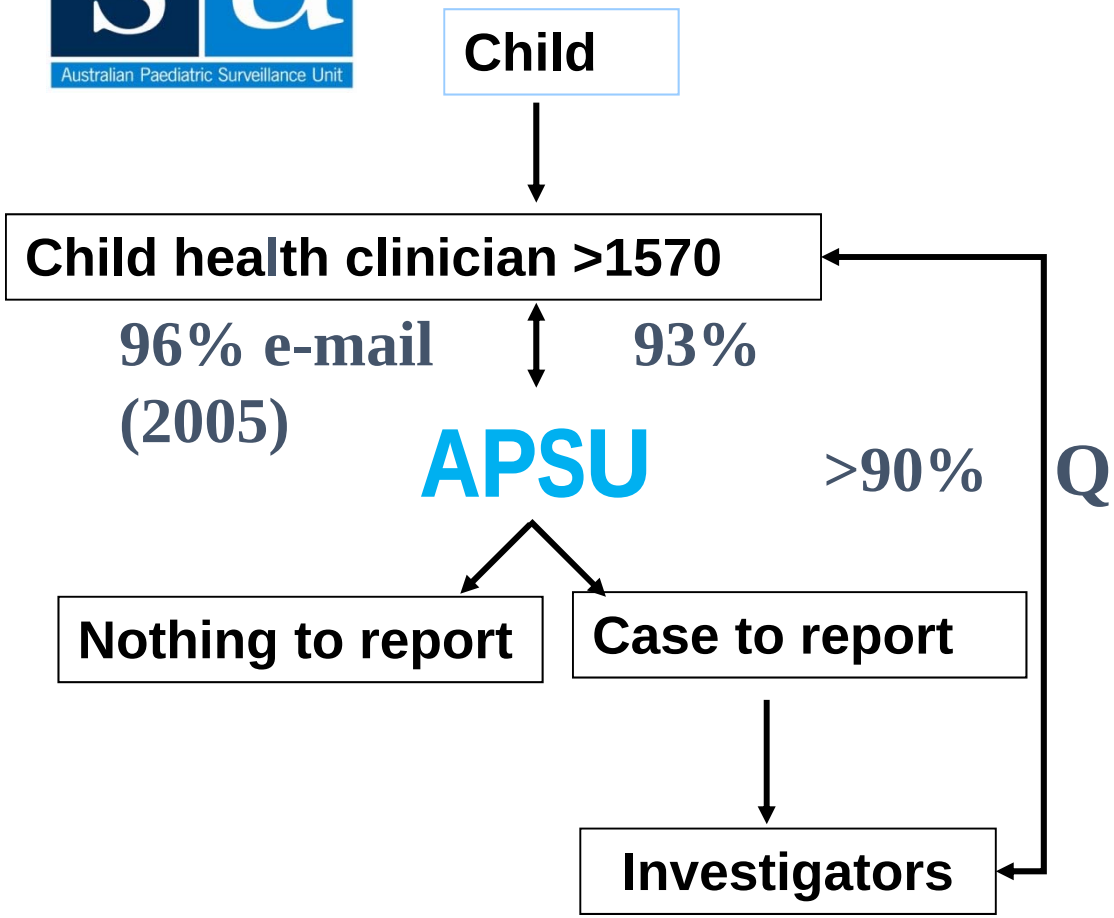


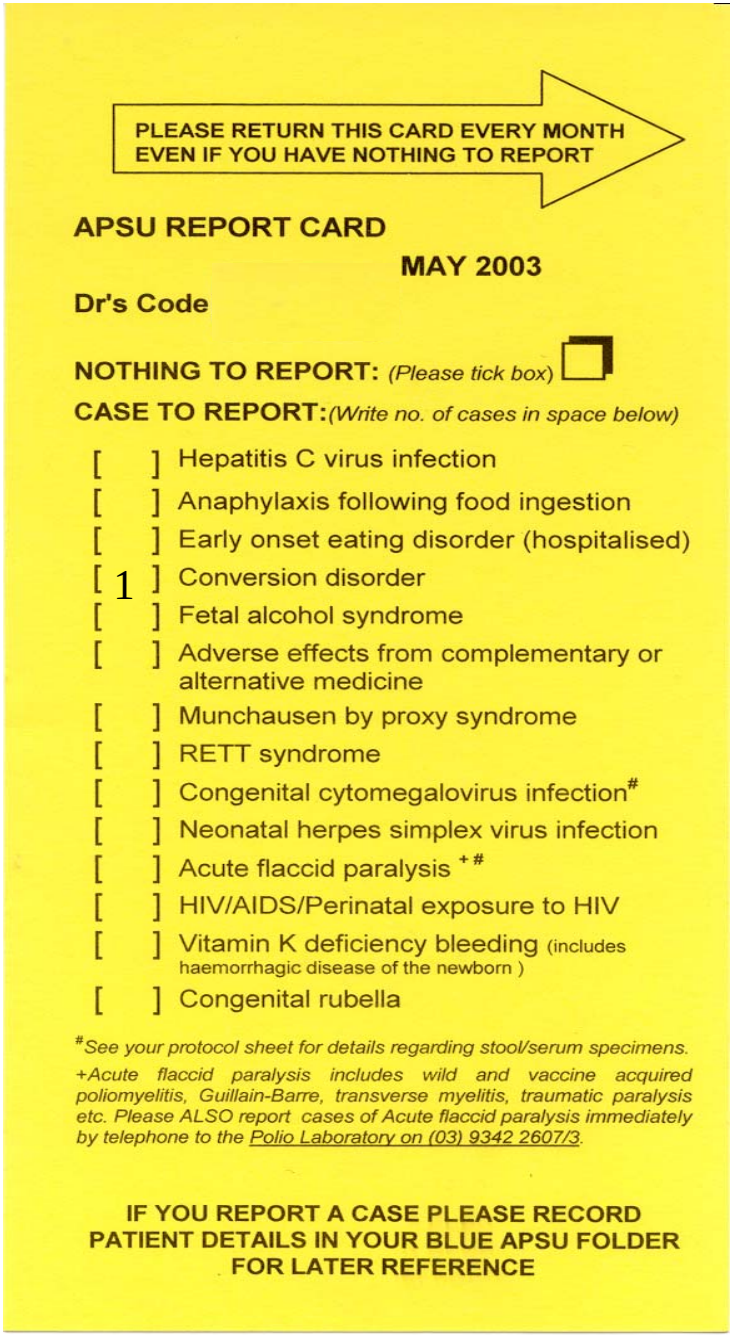
APSU – Who reports?

- ~1573 clinicians
  - 85% paediatricians, other child health specialists
  - others added as required e.g. ENT surgeons JORRP
- 4.5 million children <15y

State	Paediatricians N (%)	Population <15yrs N (%)
NSW	573 (36)	1,427,486 (32)
ACT	31 (2)	73,678 (2)
Vic	369 (23)	1,085,623 (24)
SA		299,026 (7)
WA	172 (11)	497,720 (11)
Qld	270 (17)	944,230 (21)
Tas	26 (2)	94,190 (2)
NT	20 (1)	53,943 (1)
Total	**1573**	**4,476,393**

How does APSU work?





PLEASE RETURN THIS CARD EVERY MONTH
EVEN IF YOU HAVE NOTHING TO REPORT

APSU REPORT CARD

MAY 2003

Dr's Code

NOTHING TO REPORT: *(Please tick box)* ☐

CASE TO REPORT: *(Write no. of cases in space below)*

- [ ] Hepatitis C virus infection
- [ ] Anaphylaxis following food ingestion
- [ ] Early onset eating disorder (hospitalised)
- [1] Conversion disorder
- [ ] Fetal alcohol syndrome
- [ ] Adverse effects from complementary or alternative medicine
- [ ] Munchausen by proxy syndrome
- [ ] RETT syndrome
- [ ] Congenital cytomegalovirus infection#
- [ ] Neonatal herpes simplex virus infection
- [ ] Acute flaccid paralysis +#
- [ ] HIV/AIDS/Perinatal exposure to HIV
- [ ] Vitamin K deficiency bleeding (includes haemorrhagic disease of the newborn)
- [ ] Congenital rubella

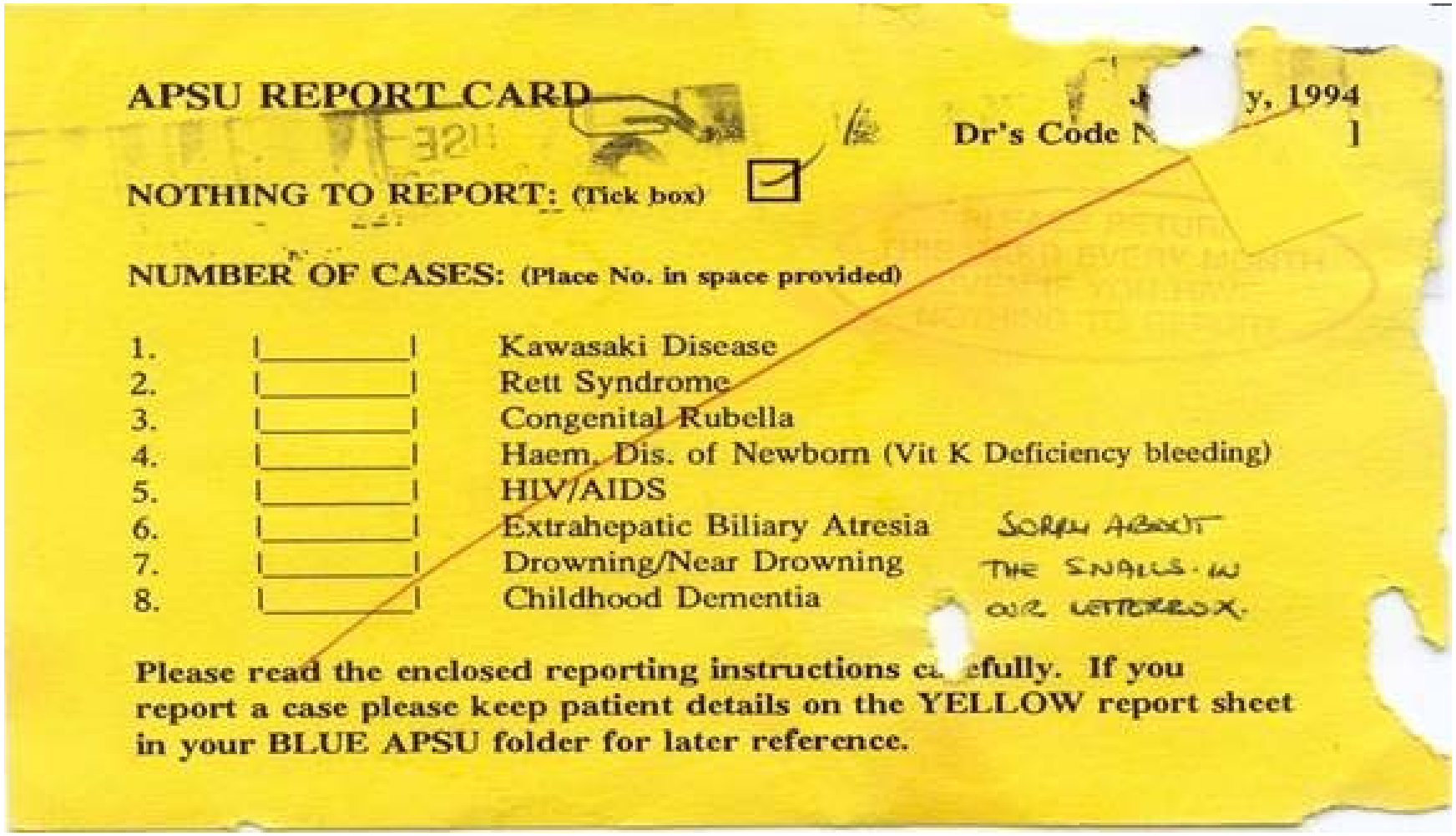
#See your protocol sheet for details regarding stool/serum specimens.

+Acute flaccid paralysis includes wild and vaccine acquired poliomyelitis, Guillain-Barre, transverse myelitis, traumatic paralysis etc. Please ALSO report cases of Acute flaccid paralysis immediately by telephone to the Polio Laboratory on (03) 9342 2607/3.

IF YOU REPORT A CASE PLEASE RECORD PATIENT DETAILS IN YOUR BLUE APSU FOLDER FOR LATER REFERENCE

- Up to 18 conditions; Protocols with Dx criteria, expert data review .

“Sorry about the snails in our letterbox”



APSU REPORT CARD — **Dr's Code N** — **y, 1994**

NOTHING TO REPORT: (Tick box) ☑

NUMBER OF CASES: (Place No. in space provided)

1.	_____	Kawasaki Disease
2.	_____	Rett Syndrome
3.	_____	Congenital Rubella
4.	_____	Haem. Dis. of Newborn (Vit K Deficiency bleeding)
5.	_____	HIV/AIDS
6.	_____	Extrahepatic Biliary Atresia
7.	_____	Drowning/Near Drowning
8.	_____	Childhood Dementia

SORRY ABOUT THE SNAILS IN OUR LETTERBOX.

Please read the enclosed reporting instructions c efully. If you report a case please keep patient details on the YELLOW report sheet in your BLUE APSU folder for later reference.

How do paediatricians report?
E-card and online questionnaire

APSU REPORT CARD DECEMBER 2016D

NOTHING TO REPORT? PLEASE HIT REPLY AND TYPE 'NTR' IN THE SUBJECT LINE OF THIS EMAIL

DO YOU HAVE A CASE TO REPORT? HIT REPLY AND TYPE THE NUMBER OF CASES IN THE SPACE PROVIDED BELOW

If you report a case, please record patient details for later reference

NEWLY DIAGNOSED CASES ONLY - *Please report cases diagnosed within study period only*
Study case report forms are available through the hyperlinks below or via the APSU website www.apsu.org.au

No of Cases	Study Case Report Forms (paper form for fax/email)	Web links for completion of Case Report
[ ]	Microcephaly in children < 12 months old	Microcephaly in children < 12 months old
[ ]	Early Onset Eating Disorder *(5 – 13 years old inclusive)*	EOED Online Questionnaire
[ ]	22q11.2 Deletion Syndrome	22q Online Questionnaire
[ ]	Fetal Alcohol Spectrum Disorders	FASD Online Questionnaire
[ ]	Childhood Interstitial Lung Disease	chILD Online Questionnaire
[ ]	*MECP2* Duplication Syndrome	
[ ]	Juvenile onset Recurrent Respiratory Papillomatosis	
[ ]	Congenital varicella	Vcon Online Questionnaire
[ ]	Neonatal varicella	Vneo Online Questionnaire
[ ]	Rett syndrome	
[ ]	Congenital cytomegalovirus infection – NSW– Other States	
[ ]	Newborn and infant herpes simplex virus infection	HSV Online Questionnaire
[ ]	Acute flaccid paralysis +#	
[ ]	Paediatric HIV infection OR perinatal exposure to HIV – Mother – Child	
[ ]	Vitamin K deficiency bleeding (includes haemorrhagic disease of the newborn)	VitK/HDN Online Questionnaire
[ ]	Congenital rubella	RUB Online Questionnaire

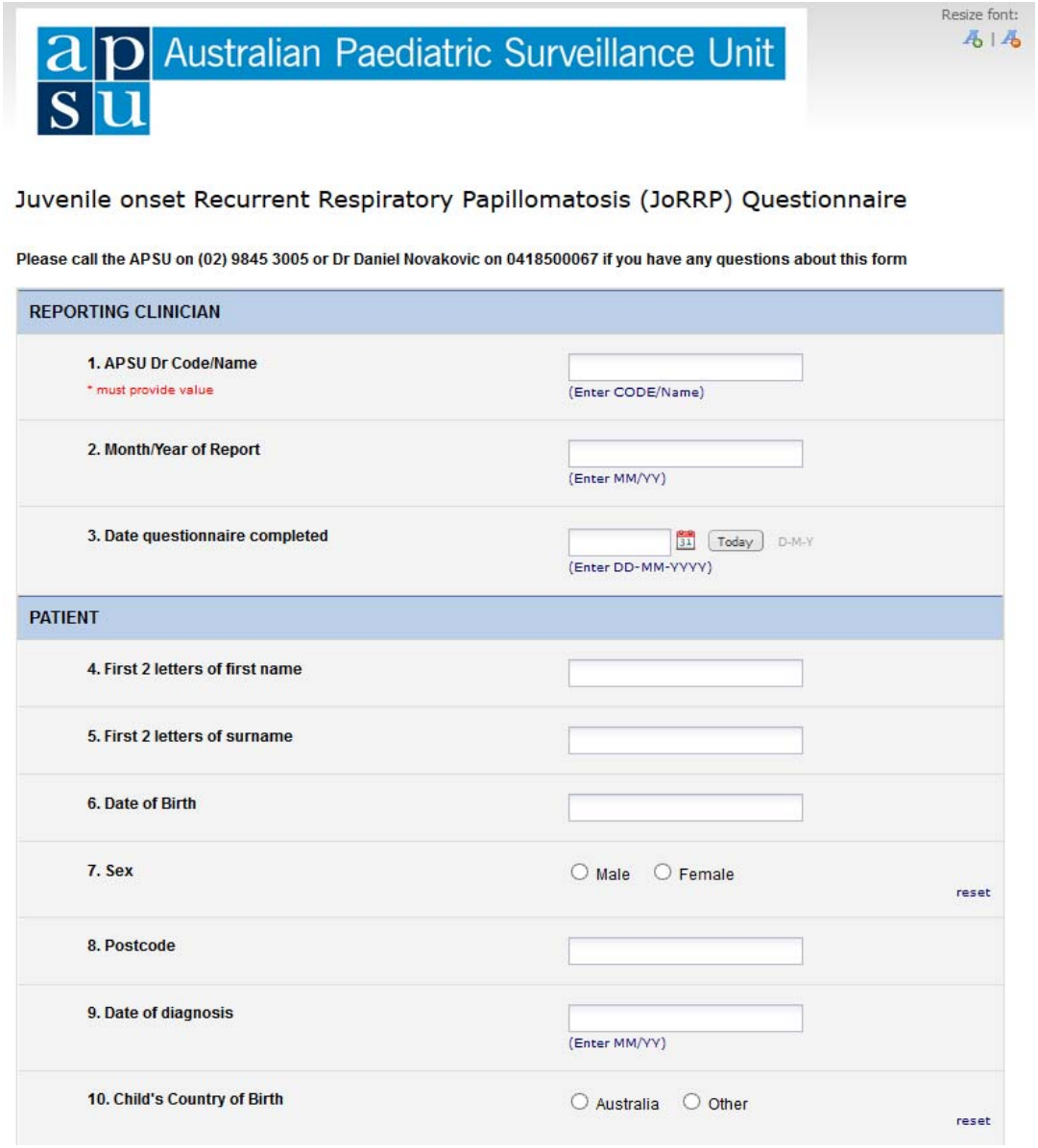
Questionnaire data downloaded

REDCap (paper)

Demographics, Dx, treatment, outcome.

Minimum identifiers collected: no names, addresses or MRN.

*FASD Register



Australian Paediatric Surveillance Unit

Resize font:

Juvenile onset Recurrent Respiratory Papillomatosis (JoRRP) Questionnaire

Please call the APSU on (02) 9845 3005 or Dr Daniel Novakovic on 0418500067 if you have any questions about this form

REPORTING CLINICIAN

1. APSU Dr Code/Name
* must provide value
(Enter CODE/Name)

2. Month/Year of Report
(Enter MM/YY)

3. Date questionnaire completed
Today D-M-Y
(Enter DD-MM-YYYY)

PATIENT

4. First 2 letters of first name

5. First 2 letters of surname

6. Date of Birth

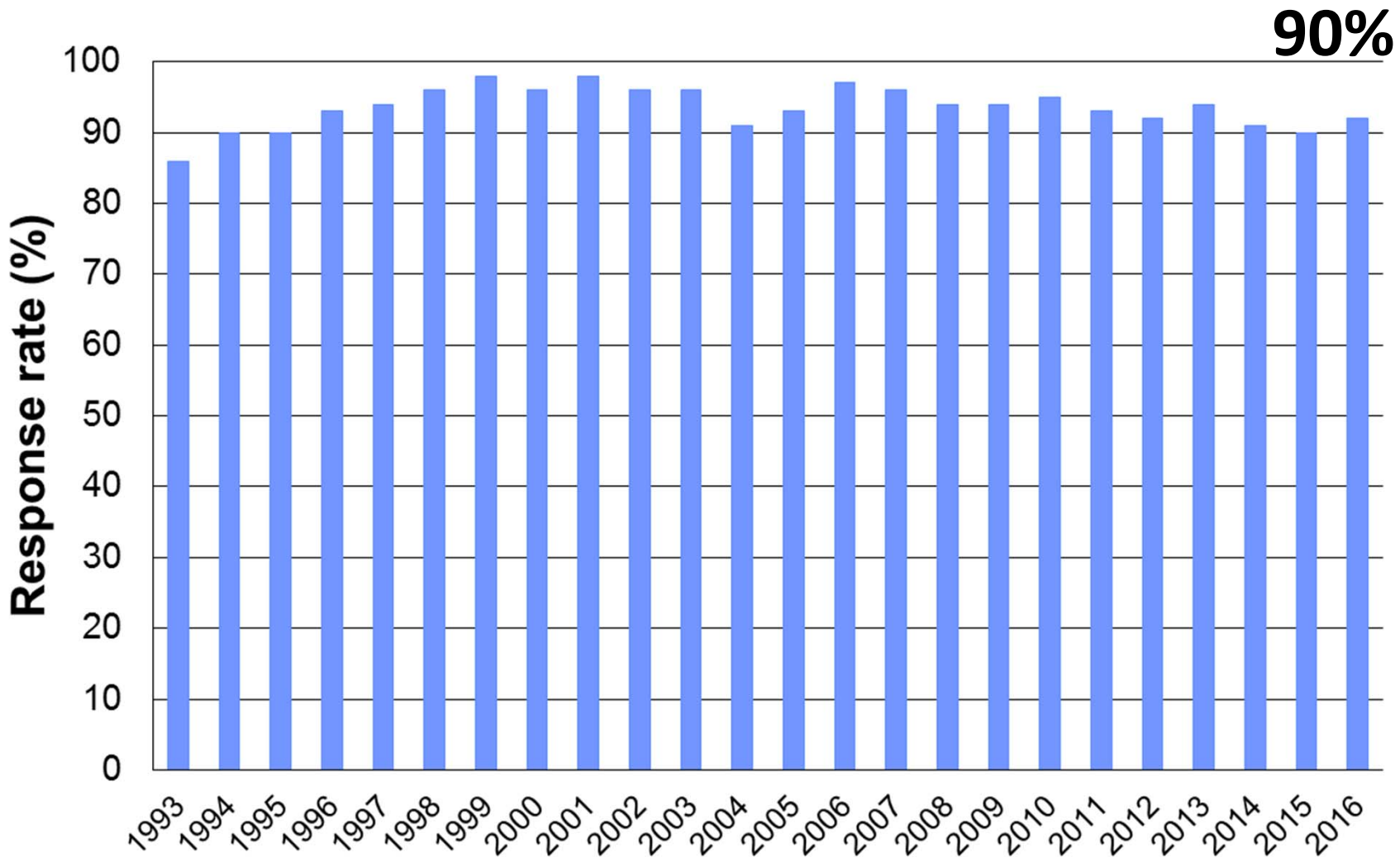
7. Sex
Male Female
reset

8. Postcode

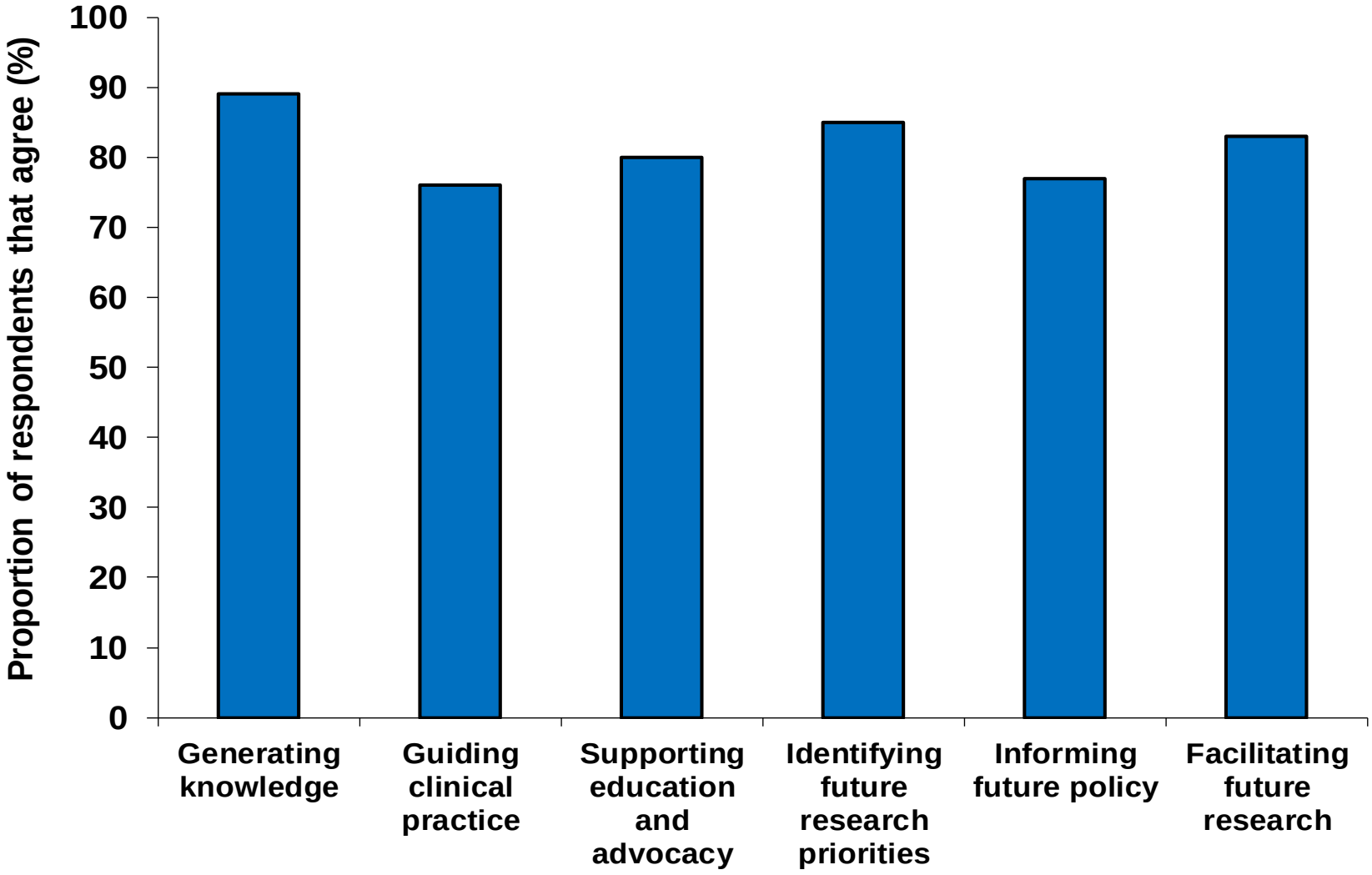
9. Date of diagnosis
(Enter MM/YY)

10. Child's Country of Birth
Australia Other
reset

Annual return rate of monthly report cards, 1993-2016



2007 Evaluation: Paediatricians (n=818)



94% monthly surveillance *not* a burden; 99% format user-friendly

Collaborating organisations

- National Centre in HIV Epidemiology and Clinical Research
- National Centre for Disease Control, DHFS
- National Centre for Immunisation Research
- National Enteric Pathogens Surveillance Lab, Melbourne
- National Perinatal Statistics Unit
- NSW Birth Defects Register
- Tropical Public Health Unit, Cairns
- SWS Area Health Authority
- Australian Polio Expert Cttee International PSUs
- AIHW
- Epidemiology Branch NSW Health Department
- WHO
- Telethon Institute, WA
- Australia, NZ Paediatric Nephrology Association
- Australian Society of Clinical Immunology and Allergy
- Victorian Infectious Diseases Reference Laboratory
- CHARGE Association
- Rett Syndrome Association
- Centre for Prevention of Psychological Problems in Children
- National Polio Reference lab

Key Facts – 25 years of surveillance

- >300 researchers, groups
- 352,944 report cards (321,179 returned)
- 68 studies; first national data
- Detailed data on ~8,330 children
- ~ 400 publications, >500 presentations, >300 media
- >90% response for 25 years

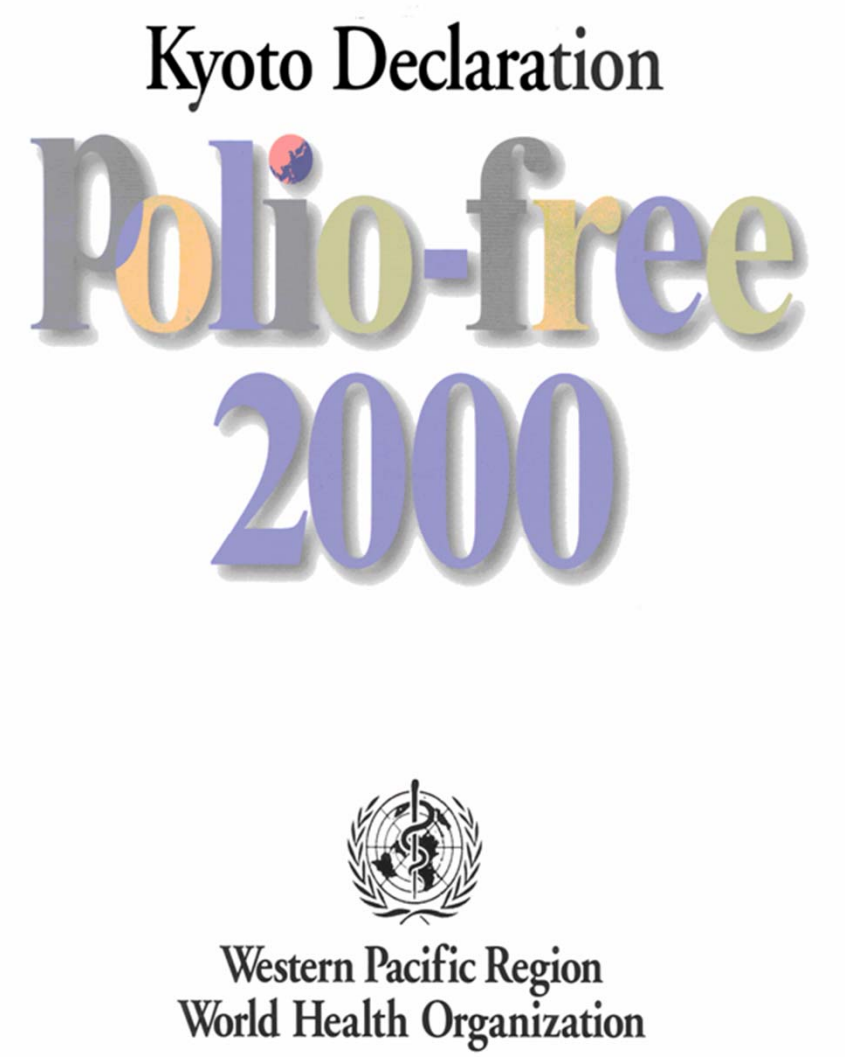
  990 clinicians in 1993; 1570 in 2017
- One-off surveys very rare conditions e.g. FGM/C
- Emergency surveillance: severe complications influenza
- Established INOPSU, PAEDS, AMOSS
- ~$20 million

TRANSLATION OF RESEARCH

AFP (1993-2018): *Peter McIntyre*. NCIRS. Contributed to WPR declared polio free in 2000

"Congratulations to the APSU for taking this initiative. Australia has undoubtedly taken the lead in the commitment to poliomyelitis eradication, as a non-endemic country, by establishing AFP surveillance"

World Health Organisation, 1995.



THE WEEKEND AUSTRALIAN JULY 28-29 2007

20 HEALTH

Polio never far away in the jet age

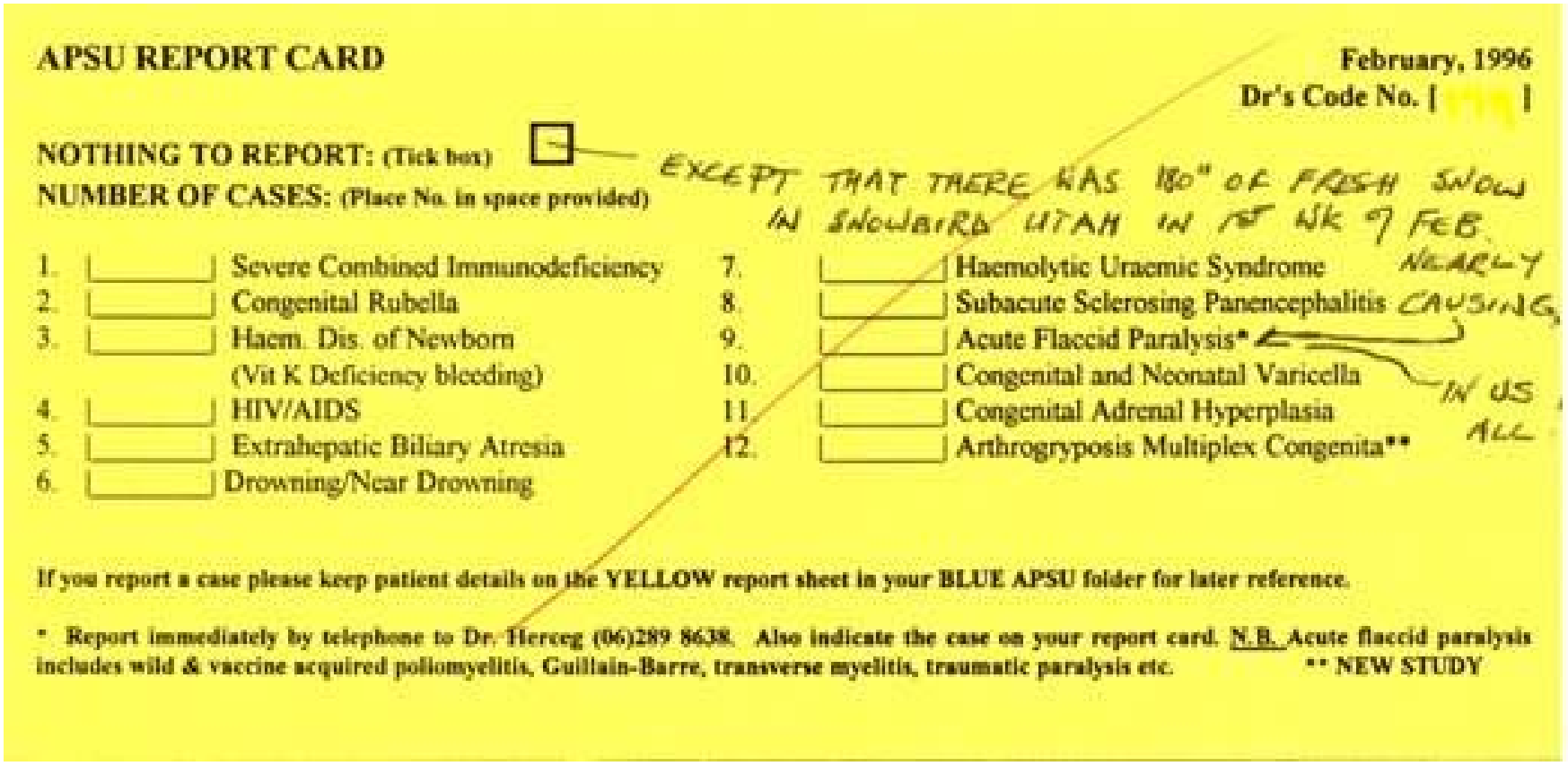
Australia's first case of polio in 21 years underlines the importance of continued vaccination. **Neena Bhandari** reports

Weekend Australia, July 2007

Need for ongoing surveillance

- 2006-7 large outbreaks in Indonesia
- 2007 case imported into Australia from Pakistan

Nothing to report “except that there was 180” of fresh snow in Snowbird in the 1st week of Feb. nearly causing acute flaccid paralysis in us all”



APSU REPORT CARD

February, 1996

Dr's Code No. [ ]

NOTHING TO REPORT: (Tick box) ☒ EXCEPT THAT THERE WAS 180" OF FRESH SNOW IN SNOWBIRD UTAH IN 1ST WK OF FEB. NEARLY CAUSING

NUMBER OF CASES: (Place No. in space provided)

1. [ ] Severe Combined Immunodeficiency
2. [ ] Congenital Rubella
3. [ ] Haem. Dis. of Newborn (Vit K Deficiency bleeding)
4. [ ] HIV/AIDS
5. [ ] Extrahepatic Biliary Atresia
6. [ ] Drowning/Near Drowning
7. [ ] Haemolytic Uraemic Syndrome
8. [ ] Subacute Sclerosing Panencephalitis
9. [ ] Acute Flaccid Paralysis* IN US ALL
10. [ ] Congenital and Neonatal Varicella
11. [ ] Congenital Adrenal Hyperplasia
12. [ ] Arthrogryposis Multiplex Congenita**

If you report a case please keep patient details on the YELLOW report sheet in your BLUE APSU folder for later reference.

*** Report immediately by telephone to Dr. Hereeg (06)289 8638. Also indicate the case on your report card. N.B. Acute flaccid paralysis includes wild & vaccine acquired poliomyelitis, Guillain-Barre, transverse myelitis, traumatic paralysis etc.** **** NEW STUDY**

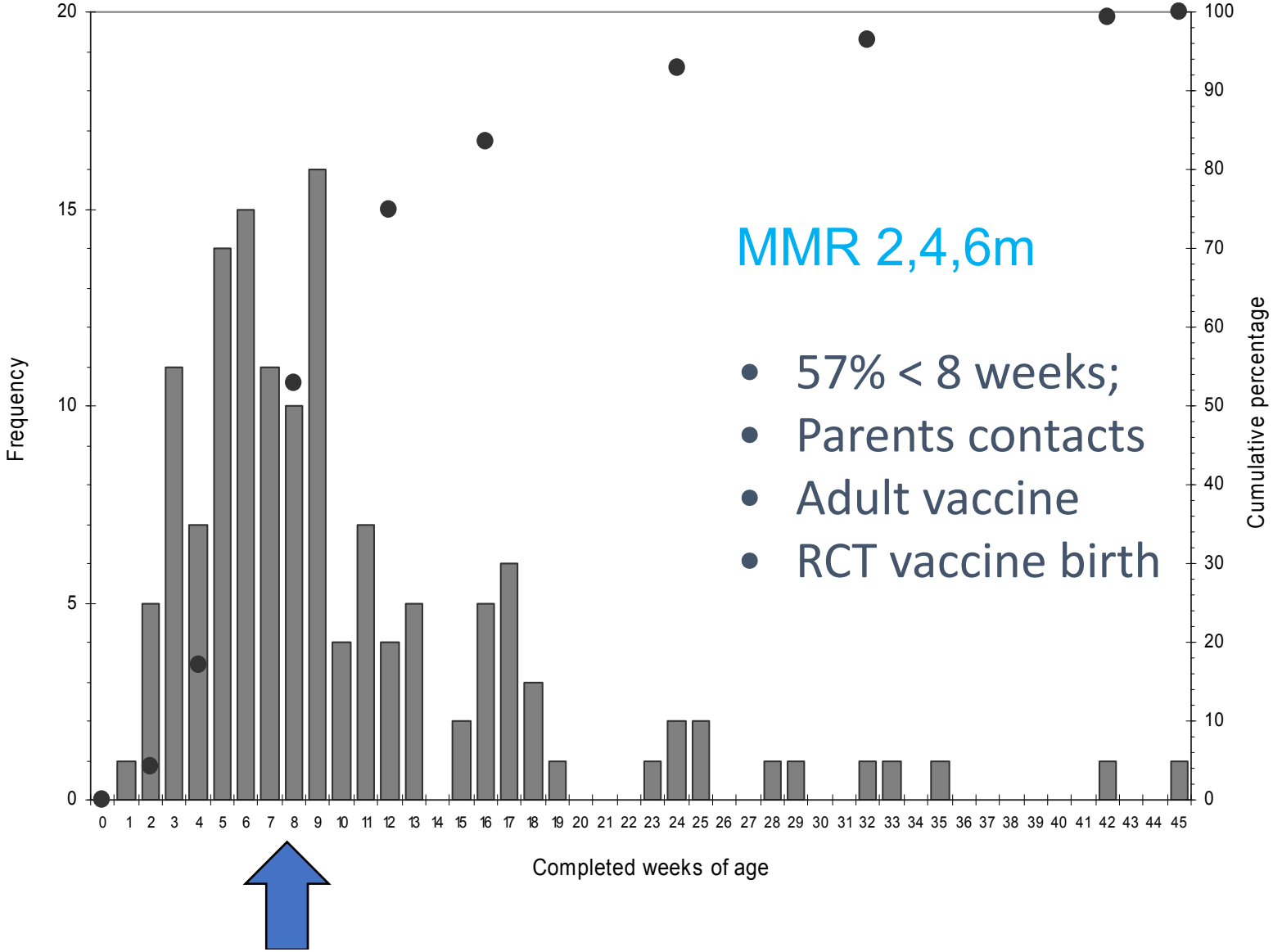
HIV infection, AIDS, Perinatal exposure HIV (1993-2018). *John Kaldor* Kirby Centre (NCHECR)

All perinatal, 85% heterosexual source, antenatal Dx, treatment, ↓transmission rates. Supported screening for all (2009).

Sydney Morning Herald: International AIDS Society, 2007

Infants hospitalised with pertussis, 2001, n=140

P McIntyre, NCIRS



Serious seat belt injuries from inappropriate or incorrectly used restraints: *Yvonne Zurynski*

Informed new laws on child restraints and seatbelts

Fetal Alcohol Syndrome (FAS) 2000.
Fetal Alcohol Spectrum Disorder (FASD) 2015

Carol Bower, Elizabeth Elliott

Two APSU studies (FAS/FASD)

NHMRC Alcohol Guidelines

WHO guidelines

RACP Alcohol policy

GP Life Scripts for Pregnancy

NHMRC funding including CRE

Australian Guide to Diagnosis

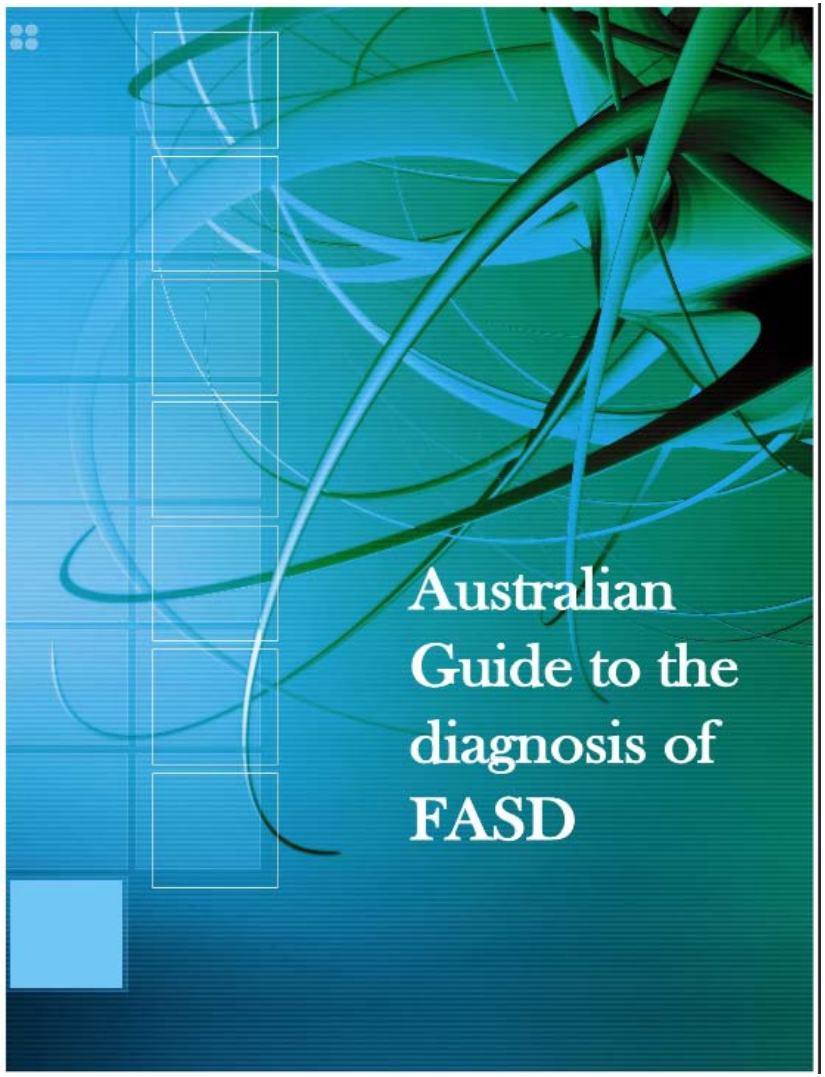
FASD Australian Registry (FASDAR)

FASD Hub Australia

HOR Inquiry FASD

Salvation Army National Awareness

Education, conferences



QUESTION

Date Wednesday, 31 March 2004
Page 22544
Questioner Allison, Sen Lyn
Speaker

Source Senate
Proof No
Responder Campbell, Sen Ian
Question No. 2588

Health: Foetal Alcohol Syndrome

Senator Allison (Victoria) asked the Minister representing the Minister for Health and Ageing, upon notice, on 25 February 2004:

What data collected by the Australian Paediatric Surv
Syndrome is available.

Senator Ian Campbell (Western Australia—Mi
Minister for Health and Ageing has provided the f

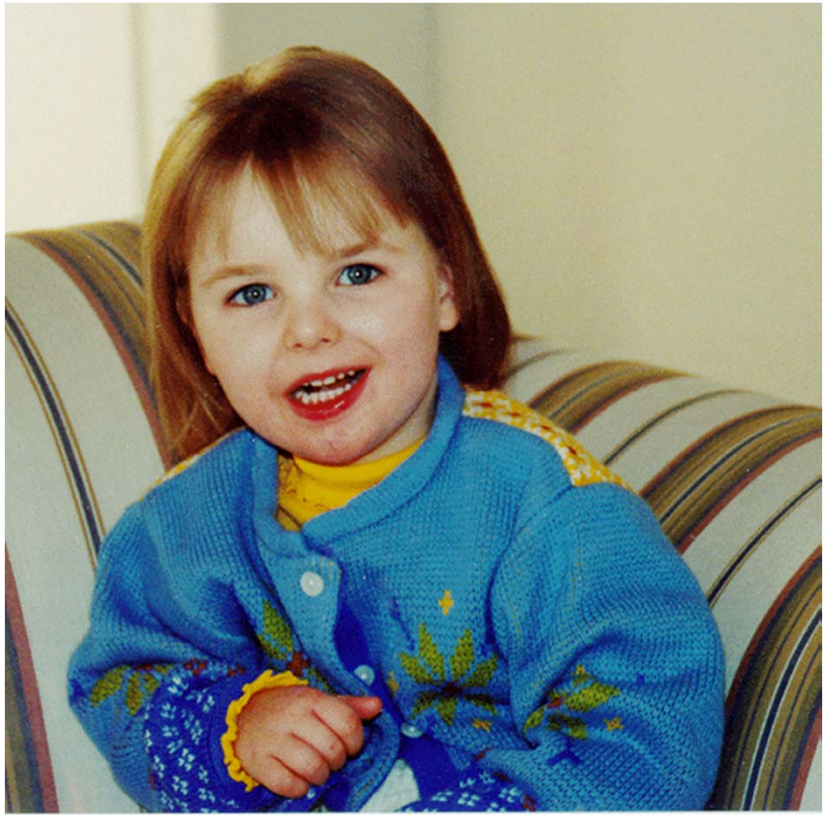
Data collected by the Australian Paediatric Surveillan
diagnoses are available by application to the Unit's direc

The APSU protocol requests clinicians working in paedi
rare childhood conditions (the study conditions). These
detailed clinical and demographic data. Study conditio
period can be extended. The study of FAS commenced i

Data collected by the Australian Paediatric Surveillance Unit (APSU) on the incidence of Foetal Alcohol Syndrome (FAS) diagnoses are available by application to the Unit's director, Associate Professor Elizabeth Elliott.

Rett Syndrome: (25 years!)

Helen Leonard



- Epidemiology
- Clinical presentation - CCS
- Genotyping
- New Genes – male encephalopathy
- Clinical severity: degree of X-inactivation
- Prognosis
- Management - RCT
- Genotype: phenotype
- NIH funding
  - Clinical characterisation (InterRett)
  - Genotyping (RettNet)
- Economic analysis
- Service development

“Rats – what happened to RETTS?”

APSU REPORT CARD May, 1995

Dr's Code No. [174]

NOTHING TO REPORT: (Tick box) ☑

NUMBER OF CASES: (Place No. in space provided)

1.		Kawasaki Disease	6.		Extrahepatic Biliary Atresia
2.		Severe Combined Immunodeficiency Syndrome	7.		Drowning/Near Drowning
			8.		Childhood Dementia
3.		Congenital Rubella	9.		Haemolytic Uraemic Syndrome
4.		Haem. Dis. of Newborn (Vit K Deficiency bleeding)	10.		Subacute Sclerosing Panencephalitis
			11.		Acute Flaccid Paralysis*
5.		HIV/AIDS	12.		Congenital and Neonatal Varicella

* Initial reporting by telephone to Dr. Herceg (06)289 8638. Please also indicate the case on your card. N.B. Acute flaccid paralysis includes wild & vaccine acquired poliomyelitis, Guillaine-Barre, transverse myelitis, traumatic paralysis etc.

Please read the enclosed reporting instructions carefully. If you report a case please keep patient details on the YELLOW report sheet in your BLUE APSU folder for later reference.

RATS – what happened to RETTS ?

Emergency surveillance: Serious complications of influenza in hospitalized children

- Following child deaths
- DoHA initiated study
- September 2007
- 10-day lead time
- Weekly surveillance
- High response rate
- Potential other emergencies (SARS, bird-flu)

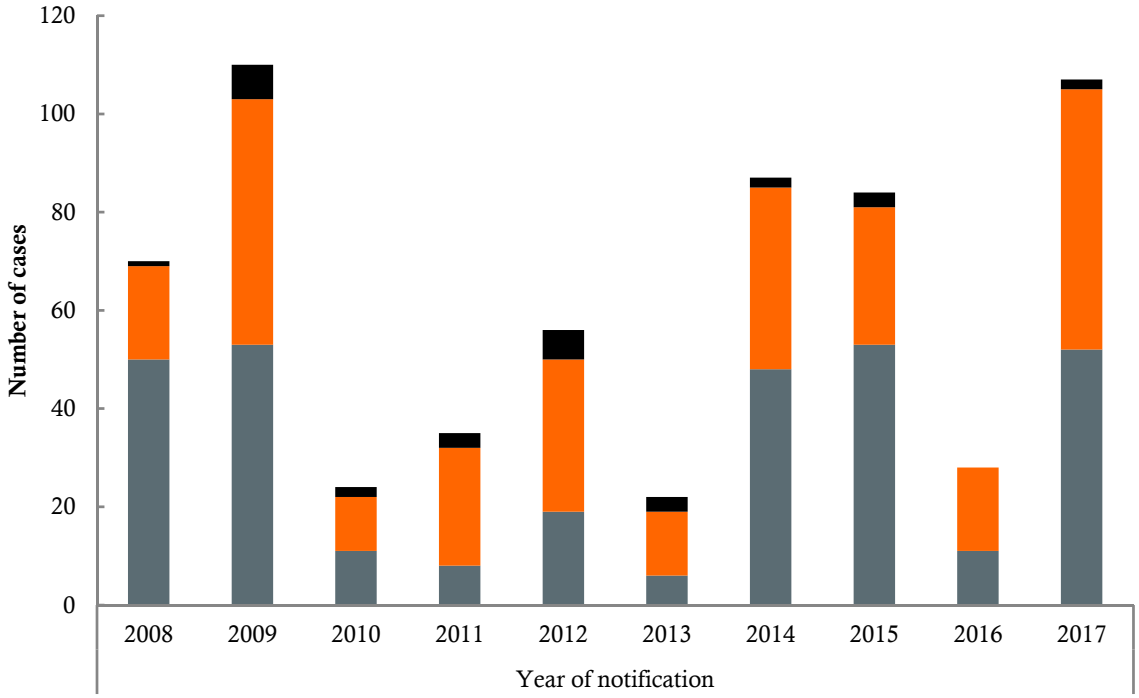
ENHANCED SURVEILLANCE FOR SERIOUS COMPLICATIONS OF INFLUENZA IN CHILDREN: ROLE OF THE AUSTRALIAN PAEDIATRIC SURVEILLANCE UNIT

Yvonne A Zurynski, David Lester-Smith, Marino S Festa, Alison M Kesson, Robert Booy, Elizabeth J Elliott

Abstract

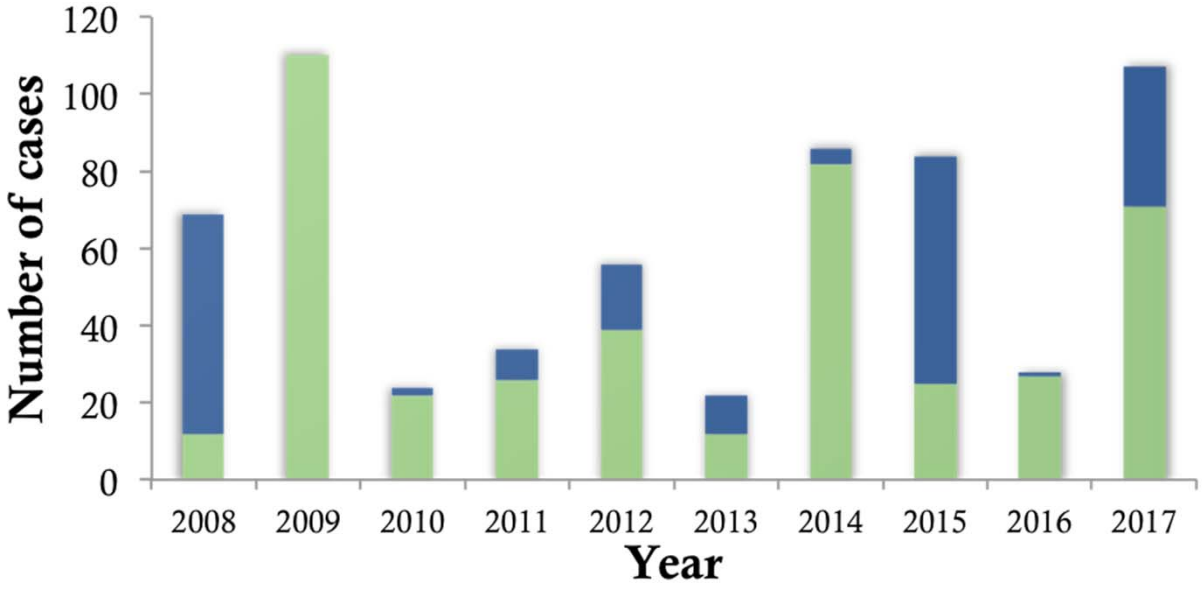
Influenza contributes significantly to disease burden among children aged less than five years. Existing influenza surveillance systems do not provide detailed data on clinical presentation, management, vaccination status, risk factors and complications in hospitalised children, or link such data with laboratory results. Following a

life-threatening multi-system complications.[1-9] Significant morbidity and mortality of influenza has been reported in Australian children, with an estimated hospitalisation rate of 82 per 100,000 and death rate of 0.2 per 100,000 children aged less than five years.[10] Of 22 children admitted with complications of influenza to one paediatric intensive care unit (PICU) over a short period in 2003, three



623 confirmed cases

- Most<2y, heathy
- 8% ATSI
- 283 (45.4%) ICU
- 9 (0-131) Bed Days
- 4.6% deaths >5y, CNS
- ~3 deaths per year

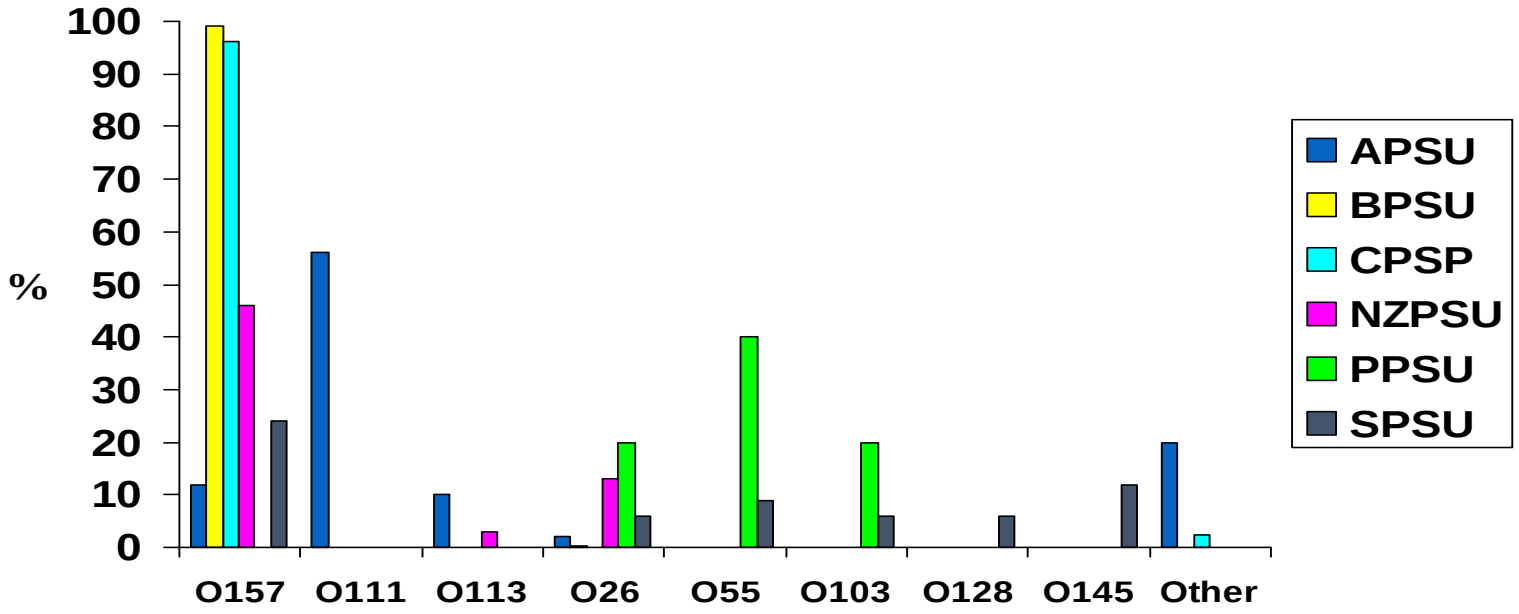


Influenza types:
426(68.4%) Flu A
194 (31.1%) Flu B
3 flu type not specified

Impacts

- **Haemolytic uremic syndrome:**
  - identified prevalent STEC in D+HUS in Australia (0111:H-)
  - revision of code of production for fermented meats
  - 10 year outcomes for atypical HUS
  - International comparison epidemiology INOPSU

Haemolytic uraemic syndrome
ST-producing isolates



*****Serotypes of STEC and Shigella dysenteriae as a proportion of all stool isolates; 50% in Australia were non-motile O157 serotype; *Shigella dysenteriae: 1 case in* Portugal, Britain & Canada *;* Britain - All but one (O26) of the STEC was O157

Other Impacts

- **EOED:**
- Severe disease <13y; 20% boys, 11y, most hospitalised, 46% unstable, 74% co-morbid psychiatric disorder
- 80% AN, 20% Avoidant Restrictive Food Intake Disorder
- Informed diagnostic criteria (DSM-5) for ED age <13 years
- international study (Canada, UK)

- **Alternative medicines:** Described dangers, mortality from inappropriate use of complementary medicines or withholding prescription medication

- **Microcephaly:** Rapid response to government-requested surveillance - potential risk of Zika Virus infection

ADC Online First, published on January 12, 2017 as 10.1136/archdischild-2016-311540

OPEN ACCESS

Female genital mutilation in children presenting to Australian paediatricians

Yvonne Zurynski,[1,2] Amy Phu,[1,2] Premala Sureshkumar,[1,2] Sarah Cherian,[3,4] Marie Deverell,[1,2] Elizabeth J Elliott,[1,2] for Australian Paediatric Surveillance Unit Female Genital Mutilation Study Steering Committee

[1]Australian Paediatric Surveillance Unit, The Children's Hospital at Westmead, Sydney, New South Wales, Australia
[2]Discipline of Child and Adolescent Health, Sydney Medical School, The University of Sydney, Sydney, New South Wales, Australia
[3]Refugee Health Service, Princess Margaret Hospital for Children, Perth, Western Australia, Australia
[4]School of Paediatrics and Child Health, University of Western Australia, Perth, Western Australia, Australia

Correspondence to
Dr Yvonne Zurynski, The Australian Paediatric Surveillance Unit, The Children's Hospital at Westmead, Locked Bag 4001, Westmead, Sydney, NSW 2145, Australia; yvonne.zurynski@health.nsw.gov.au

Received 30 June 2016
Revised 4 October 2016
Accepted 27 November 2016

ABSTRACT

Objective The WHO reports that female genital mutilation/cutting (FGM/C) is an ancient cultural practice prevalent in many countries. FGM/C has been reported among women resident in Australia. Our paper provides the first description of FGM/C in Australian children.

Design Cross-sectional survey conducted in April–June 2014.

Setting Paediatricians and other child health specialists recruited through the Australian Paediatric Surveillance Unit were asked to report children aged <18 years with FGM/C seen in the last 5 years, and to provide data for demographics, FGM/C type, complications and referral for each case.

Participants Of 1311 eligible paediatricians/child health specialists, 1003 (76.5%) responded.

Results Twenty-three (2.3%) respondents had seen 59 children with FGM/C and provided detailed data for 31. Most (89.7%) were identified during refugee screening and were born in Africa. Three (10.3%) were born in Australia: two had FGM/C in Australia and one in Indonesia. All parents were born overseas, mainly Africa (98.1%). Ten children had WHO FGM/C type I, five type II, five type III and six type IV. Complications in eight children included recurrent genitourinary infections, menstrual, sexual, fertility and psychological problems. Nineteen children (82.6%) were referred to obstetrics/gynaecology: 16 (69.9%) to social work and 13 (56.5%) to child protection.

Conclusions This study confirms that FGM/C is seen in paediatric clinical practice within Australia. Paediatricians need cultural awareness, education and resources to help them identify children with FGM/C and/or at risk of FGM/C, to enable appropriate referral and counselling of children, families and communities to assist in the prevention of this practice.

What is already known on this topic?

- UNICEF estimates that 200 million girls and women have undergone female genital mutilation/cutting (FGM/C) and the procedure is usually done in girls aged 1 month to 15 years.
- FGM/C has been reported among immigrant women living in developed countries.
- There is only one report of FGM/C in children living in developed countries—a study of children in the UK showed that type IV FGM/C was the most common.

What this study adds?

- Our paper provides the first report of FGM/C in Australian children.
- All four types of FGM/C were seen in the children identified by our study, with type I the most common.
- Our study highlights that child health services need to be included in any response to end the practice of FGM/C and child health professionals need education, resources and referral pathways for children with FGM/C.

without removal of the clitoris.[3 4] A classification system developed by the WHO identifies four distinct types of FGM/C (figure 1).[5 6] FGM/C violates the United Nations (UN) Charter of Human Rights, the UN Charter of Women's Rights, the

Juvenille Onset Recurrent Respiratory Papillomatosis (JORRP)
Vertical transmission, focal activation of HPV types 6 and 11

The Journal of Infectious Diseases

MAJOR ARTICLE

A Prospective Study of the Incidence of Juvenile-Onset Recurrent Respiratory Papillomatosis After Implementation of a National HPV Vaccination Program

Daniel Novakovic,[1] Alan T. L. Cheng,[1,2] Yvonne Zurynski,[1,3] Robert Booy,[4] Paul J. Walker,[5] Robert Berkowitz,[6] Henley Harrison,[7] Robert Black,[8] Christopher Perry,[8] Shyan Vijayasekaran,[9] David Wabnitz,[10] Hannah Burns,[8] Sepehr N. Tabrizi,[11,12] Suzanne M. Garland,[11,12] Elizabeth Elliott,[3] and Julia M. L. Brotherton[13,14]

[1]University of Sydney Medical School; [2]ENT Department, Children's Hospital Westmead; [3]Australian Paediatric Surveillance Unit, University of Sydney and Kids Research Institute, Sydney Children's Hospitals Network (Westmead); [4]National Centre for Immunisation Research and Surveillance, Children's Hospital Westmead; [5]Otolaryngology Department, John Hunter Children's Hospital, Newcastle, New South Wales; [6]Otolaryngology Department, Royal Children's Hospital, Melbourne, Victoria; [7]ENT Department, Sydney Children's Hospital, Randwick, New South Wales; [8]Paediatric Otolaryngology Head and Neck Surgery Department, Lady Cilento Children's Hospital, Brisbane, Queensland; [9]Department of Otolaryngology - Head and Neck Surgery, Princess Margaret Hospital for Children, Perth, Western Australia; [10]Department of Otolaryngology - Head and Neck Surgery, Women's and Children's Hospital, Adelaide, South Australia; [11]Royal Women's Hospital, Department of Microbiology and Infectious Diseases, and Murdoch Children's Research Institute, Infection and Immunity Theme, Parkville; [12]Department of Obstetrics and Gynaecology, University of Melbourne, Parkville; [13]National HPV Vaccination Program Register, VCS, East Melbourne; and [14]School of Population and Global Health, University of Melbourne, Parkville, Victoria, Australia

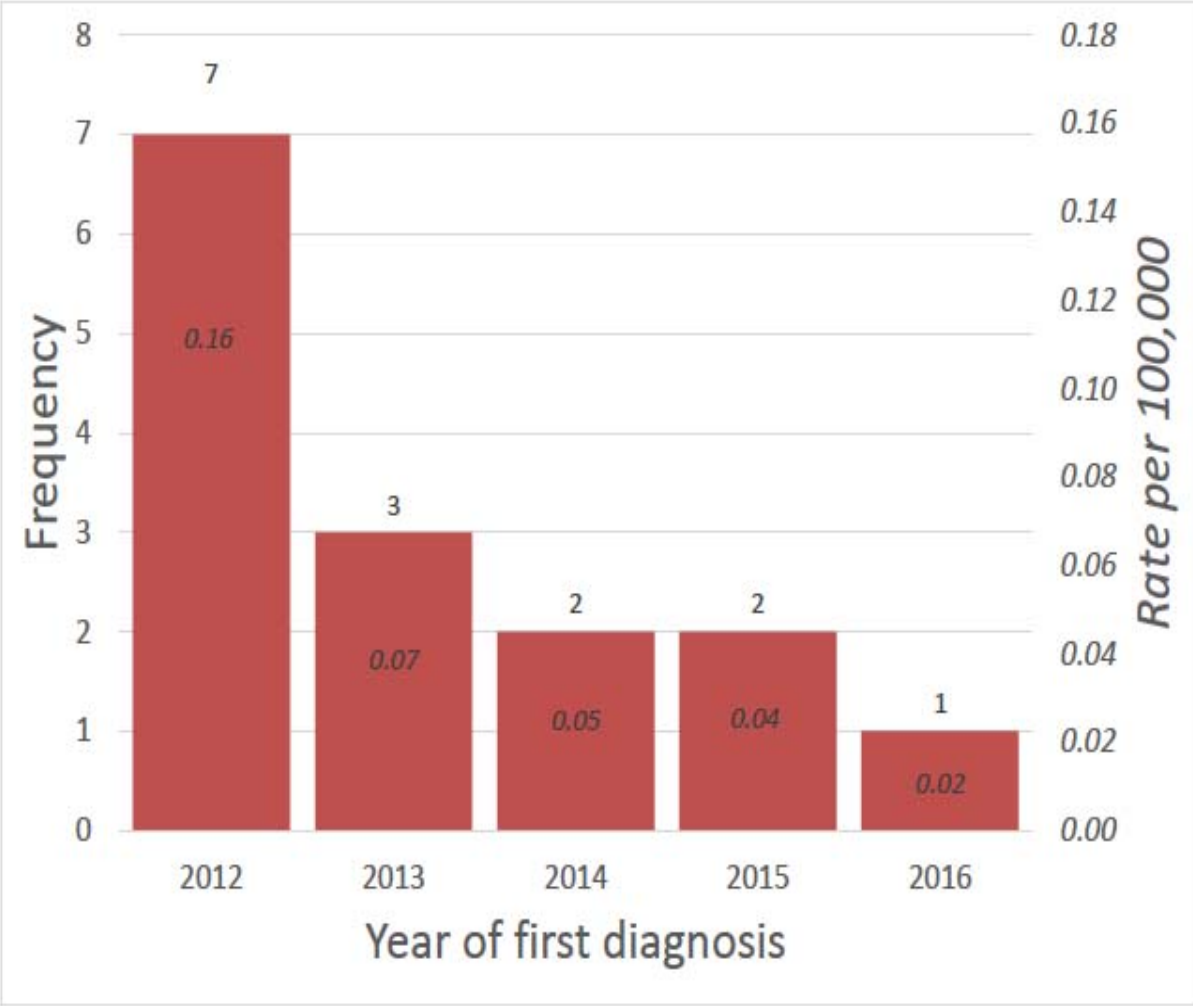
Background. Recurrent respiratory papillomatosis is a rare but morbid disease caused by human papillomavirus (HPV) types 6 and 11. Infection is preventable through HPV vaccination. Following an extensive quadrivalent HPV vaccination program (females 12–26 years in 2007–2009) in Australia, we established a method to monitor incidence and demographics of juvenile-onset recurrent respiratory papillomatosis (JORRP) cases.

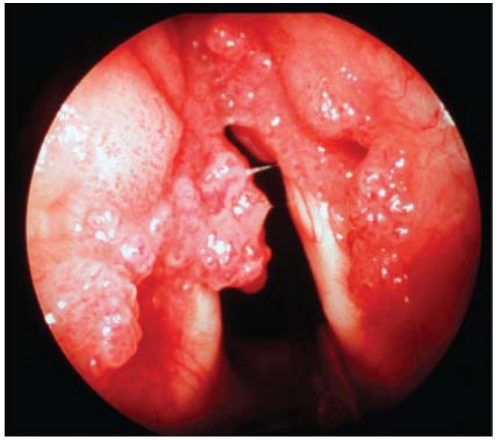
Methods. The Australian Paediatric Surveillance Unit undertakes surveillance of rare pediatric diseases by contacting practitioners monthly. We enrolled pediatric otorhinolaryngologists and offered HPV typing. We report findings for 5 years to end 2016.

Results. The average annual incidence rate was 0.07 per 100 000. The largest number of cases was reported in the first year, with decreasing annual frequency thereafter. Rates declined from 0.16 per 100 000 in 2012 to 0.02 per 100 000 in 2016 ($P = .034$). Among the 15 incident cases (60% male), no mothers were vaccinated prepregnancy, 20% had maternal history of genital warts, and 60% were first born; 13/15 were born vaginally. Genotyped cases were HPV-6 (n = 4) or HPV-11 (n = 3).

Conclusion. To our knowledge, this is the first report internationally documenting decline in JORRP incidence in children following a quadrivalent HPV vaccination program.

Decreasing incidence 2012-2016: effectiveness of HPV vaccine program





- Average incidence 0.07 per 100,000 pa
- Significant decrease 0.16/100,000 in 2012 to 0.02/100,000 in 2016 (P=0.03)

Brotherton et al. J Infect Dis 2018;217:1504–5

Guidelines Prevention, Management of Sudden Unexpected Death in Infancy <7 days (n=48)

Prof Heather Jeffery

National: NHMRC Guidelines – draft form

State:

2012 Maternity-Safer sleeping practices for babies in NSW

2008 Death - Management of SUDI

Hospital:

Prevention first

What to do if SUDI/Collapse occurs (RPA Protocol on web)

Importance of perinatal post mortem by competent, relevant pathologist (perintal or paediatric pathologist)

Discharge planning

Early childhood centres

Community awareness

SUPPORTING NEW SURVEILLANCE SYSTEMS

International Network of Paediatric Surveillance Units, 1998
Foster International collaboration

Paediatr Child Health. 2001 May;6(5):251-60.

Rare disease surveillance: An international perspective.

Elliott EJ, Nicoll A, Lynn R, Marchessault V, Hirasing R, Ridley G.

University of Sydney, Royal Alexandra Hospital for Children, Sydney, Australia.

Abstract

BACKGROUND: The International Network of Paediatric Surveillance Units (INoPSU) Ottawa, Ontario in June 2000.

- **>50 mill <15y**
- **>400 studies**

INOPSU

Member Units

Australian Paediatric Surveillance Unit (APSU)*

Belgium Paediatric Surveillance Unit

British Paediatric Surveillance Unit (BPSU)*

Canadian Paediatric Surveillance Program (CPSP)*

German Paediatric Surveillance Unit (ESPED)*

Irish Paediatric Surveillance Unit (IPSU)

Netherlands Paediatric Surveillance Unit (NSCK)*

New Zealand Paediatric Surveillance Unit (NZPSU)*

Portugal Paediatric Surveillance Unit (PPSU)

Swiss Paediatric Surveillance Unit (SPSU)*

Welsh Paediatric Surveillance Unit (WPSU)

**Indicates inaugural units who formed INoPSU*

Australian Maternity Outcomes Surveillance System (AMOSS)

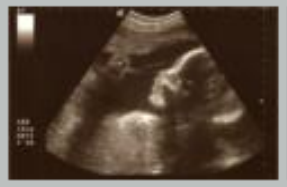
AMOSS
ralasian Maternity Outcomes Surveillance System

The Australasian Maternity Outcomes Surveillance System (AMOSS) known as 'AMOSS' is a national surveillance mechanism designed to study a variety of rare or serious conditions in pregnancy, childbirth and the post natal phase. Through translating the findings from these studies into reliable evidence-based practice, the aim of AMOSS is to improve the safety and quality of maternity care in Australia and New Zealand.

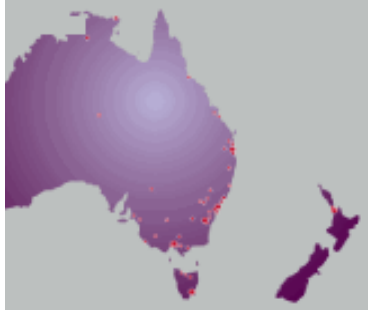
- AMOSS: its role and objectives more >
- How AMOSS works more >

TAKE A CLOSER LOOK







HOW TO GET INVOLVED

Map showing the hospitals where AMOSS data collection has commenced. To register your interest contact the AMOSS team or call (02) 9382 1068.

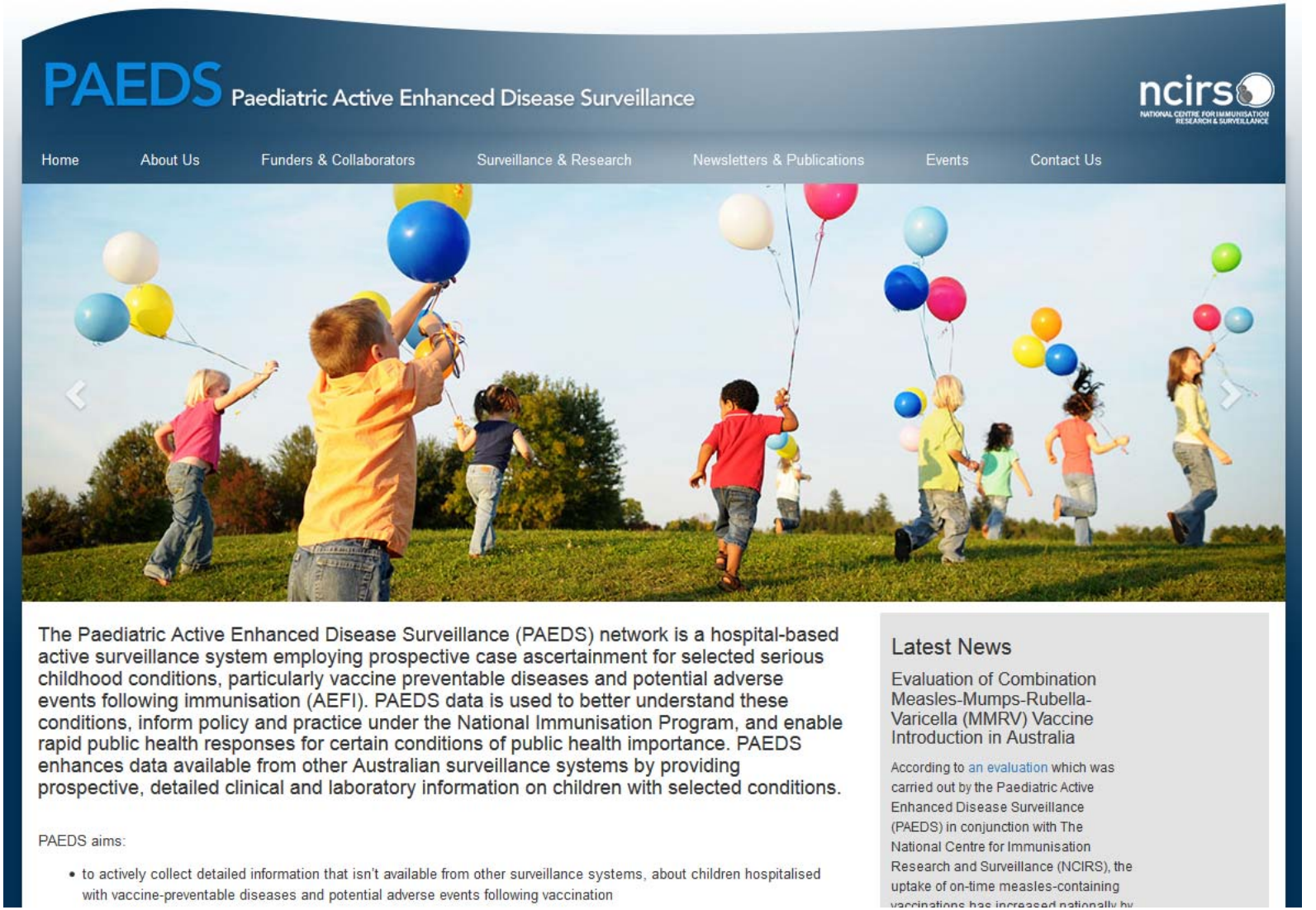
23 February 2011: Influenza in pregnancy (ICU) study ends

AMOSS has ceased data collection on influenza in pregnancy with ICU admission only. This study, an extension of the 2009 collaboration with the ANZICS INFINITE Flu registry rese... more >

Australian Government
National Health and Medical Research Council

UNSW
THE UNIVERSITY OF NEW SOUTH

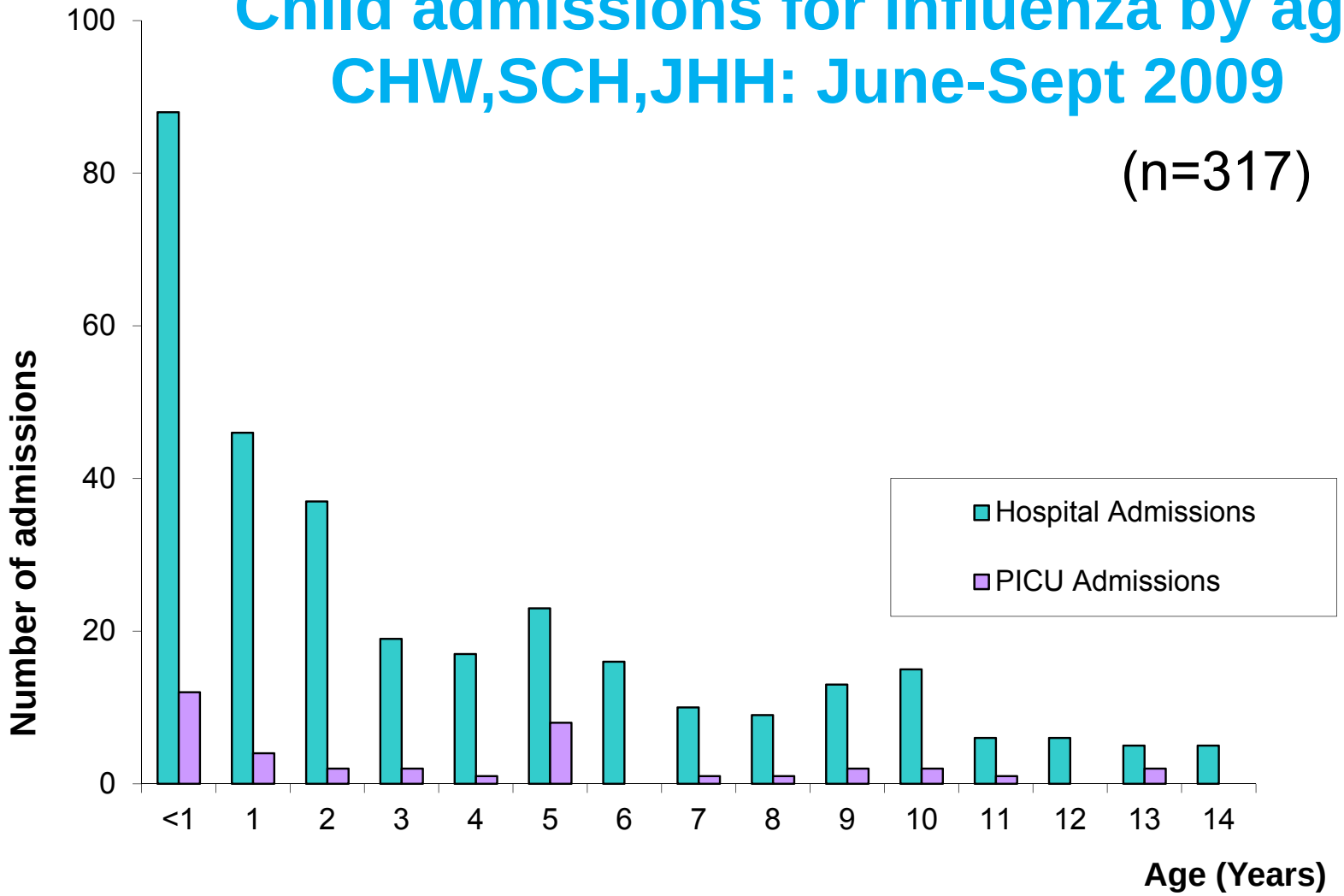
inatal & Reproductive Epidemiology Research Unit (PRERU), School of Women's & Children's Health - UNSW -



In-Patient Surveillance System: Active case-finding by nurses. Developed by APSU and NCIRS (initially funded APSU NHMRC Enabling Grant)

Child admissions for influenza by age CHW,SCH,JHH: June-Sept 2009

(n=317)



6 months: 55 (17.3%); <12 months: 84 (26.4 %)
10% PICU, O ECMO; 35% severe complications.

84% Flu A, H1N1. 6% vaccinated, 84% age-eligible, 48% chronic disease

NHMRC ‘10 of the best’ 2013
H1N1 Influenza 2009. APSU/PAEDS

Rapid response: Awareness of severe outcomes (neurological) in previously well children; antiviral underuse; need seasonal vaccination.

Contents lists available at ScienceDirect

Vaccine

journal homepage: www.elsevier.com/locate/vaccine

Intussusception following rotavirus vaccine administration: Post-marketing surveillance in the National Immunization Program in Australia

J.P. Buttery[a,b,c,d,*,1], M.H. Danchin[a,b,c,e,**,1], K.J. Lee[e,f], J.B. Carlin[e,f], P.B. McIntyre[g], E.J. Elliott[h], R. Booy[g], J.E. Bines[b,e,i], for the PAEDS/APSU Study Group

[a] SAEFVIC in Child and Adolescent Immunisation, Murdoch Children's Research Institute, Australia
[b] NHMRC CCRE in Child and Adolescent Immunisation, Murdoch Children's Research Institute, Australia
[c] Department of General Medicine, Royal Children's Hospital Melbourne, Australia
[d] Infectious Diseases Unit, Department of Paediatrics, Monash Children's Hospital, Monash University, Melbourne, Australia
[e] Department of Paediatrics, University of Melbourne, Australia
[f] Clinical Epidemiology and Biostatistics Unit, Murdoch Children's Research Institute, Australia
[g] National Centre for Immunisation Research and Surveillance, Australia
[h] Discipline of Paediatrics and Child Health, Sydney Medical School, Australian Paediatric Surveillance Unit, The Children's Hospital at Westmead and the University of Sydney, Sy
Australia
[i] Department of Gastroenterology and Clinical Nutrition, Royal Children's Hospital, Melbourne, Australia

Joint APSU/PAEDS data: Increase in intussusception detected after first dose of Rotateq vaccine investigated by Therapeutic Drugs Administration and prompted manufacturer's warning

APSU – is more than surveillance

- Audit e.g. microcephaly 10 years
- Systematic reviews/rapid reviews
- One-off study: FGM/C
- Emergency surveillance: severe flu, microcephaly
- Health services evaluation – utility, cost e.g. transition, integrated care, metabolic disease
- Impacts on families e.g. out-of-pocket expenses
- Guidelines
- Registries e.g. FASD
- Policy
- Advocacy
- Education
- Family support
- National and government committees
- Data linkage e.g. NDIS NSW

Chronic Disease Team (DoHA)

Rare Chronic Diseases in Childhood: incidence, impacts on health services and needs of families

- Children's Interstitial Lung Disease
- Non-Cystic Fibrosis Bronchiectasis
- Obesity Hypoventilation Syndrome
- Fetal Alcohol Spectrum Disorder

Transition to adult services

- Mapping, audit of services
- Workshops for adolescents
- Neuromuscular diseases

Female Genital Mutilation

- Knowledge, attitudes
- Incidence/prevalence

Impacts of Rare diseases: ARC Linkage Grant (Zurynski, Elliott, Christodoulu, Leonard)

- **Children and families:** Steve Waugh Foundation
  SMILE Foundation
  AGSA
- **Health professionals:** RACP, Paediatrics Child Health
- **Health Services:** Children's Hospital Westmead
  Western Sydney Genetics
  Management Support & Analysis Unit

Early onset, chronic complex, need impact data to inform services, policy. Ist internationally

"Going into bat for rare diseases" through parent support grants, advocacy, awareness of rare disease

Steve Waugh Foundation

Diagnosis

- 37% delayed Dx
- 27% wrong Dx
- 37% disease progression
- 25% delayed treatment
- 10% inappropriate Rx

- 69% lack HP knowledge
- 21% lack recognition symptoms
- 18% accessing testing

- 54% frustration/stress

Zurynski et al. Orphanet Journal of Rare Diseases (2017) 12:68
DOI 10.1186/s13023-017-0622-4

Orphanet Journal of Rare Diseases

RESEARCH Open Access

Australian children living with rare diseases: experiences of diagnosis and perceived consequences of diagnostic delays

Yvonne Zurynski[1,2*], Marie Deverell[1,2], Troy Dalkeith[1,3], Sandra Johnson[2], John Christodoulou[4,5,6,7], Helen Leonard[8], Elizabeth J Elliott[1,2] and APSU Rare Diseases Impacts on Families Study group

Abstract

Background: Children and families living with rare disease often experience significant health, psychosocial, economic burdens and diagnostic delays. Experiences appear to be constant, regardless of the specific rare disease diagnosis. Systematically collected Australian data to support policy response on rare diseases are scarce. We address this gap by providing survey results about 462 children aged <19 years living with approximately 200 different rare diseases.

Results: Of 462 children, 96% were born in Australia, 55% were male, median age was 8.9 years (0–18.2). Four-hundred-and-twenty-eight (93%) had received a definitive diagnosis but 29 (7%) remained undiagnosed. Before receiving the correct diagnosis 38% consulted ≥ 6 different doctors. Among those with a diagnosis, 37% believed the diagnosis was delayed and 27% initially received a wrong diagnosis. Consequences of delayed diagnosis include anxiety, loss of reproductive confidence because of an ill-defined genetic risk, frustration and stress (54%), disease progression (37%), delays in treatment (25%) and inappropriate treatments (10%). Perceived reasons for diagnostic delays included lack of knowledge about the disease among health professionals (69.2%), lack of symptom awareness by the family (21.2%) and difficulties accessing tests (17.9%). Children with inborn errors of metabolism were less likely to have a delayed diagnosis compared with other disease groups (Chi-Sq = 17.1; $P < 0.0001$), most likely due to well-established and accessible biochemical screening processes. Diagnosis was given in person in 74% of cases, telephone in 18.5% and via a letter in 3.5%. Some families (16%) were dissatisfied with the way the diagnosis was delivered, citing lack of empathy and lack of information from health professionals. Psychological support at diagnosis was provided to 47.5%, but 86.2% believed that it should always be provided. Although 74.9% of parents believed that the diagnosis could have an impact on future family planning, only 44.8% received genetic counselling.

Diagnosis can be distressing

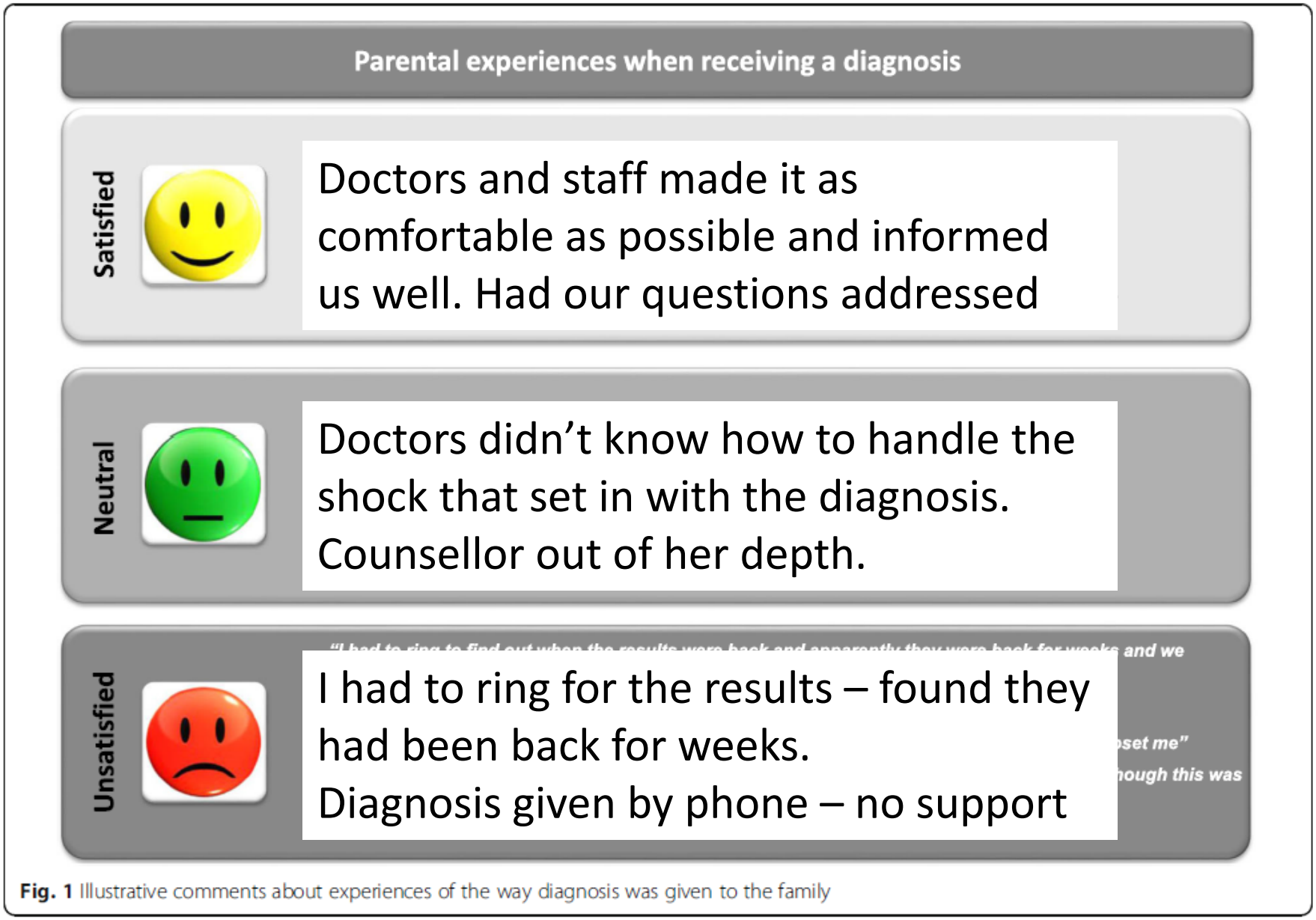


Fig. 1 Illustrative comments about experiences of the way diagnosis was given to the family

High use of health services (n=463; 200 diseases)

In one year:

- 647 ED
- 1200 admissions
- 3163 GP
- 3540 specialists
- >10000 allied health

Cost estimate ED presentations, admissions:

$8,771,082 p.a. or $25,277 p.a. per child

One child with CHILD to 8 years ~$1mill

Barriers to accessing health services, 43%

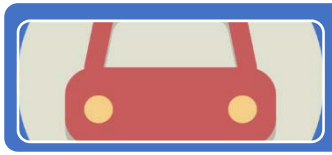
More frequent in regional/remote areas $p<0.0001$


Out of pocket costs


Care for siblings


Long waiting times


Time away from employment


Long distance to travel


Services not available

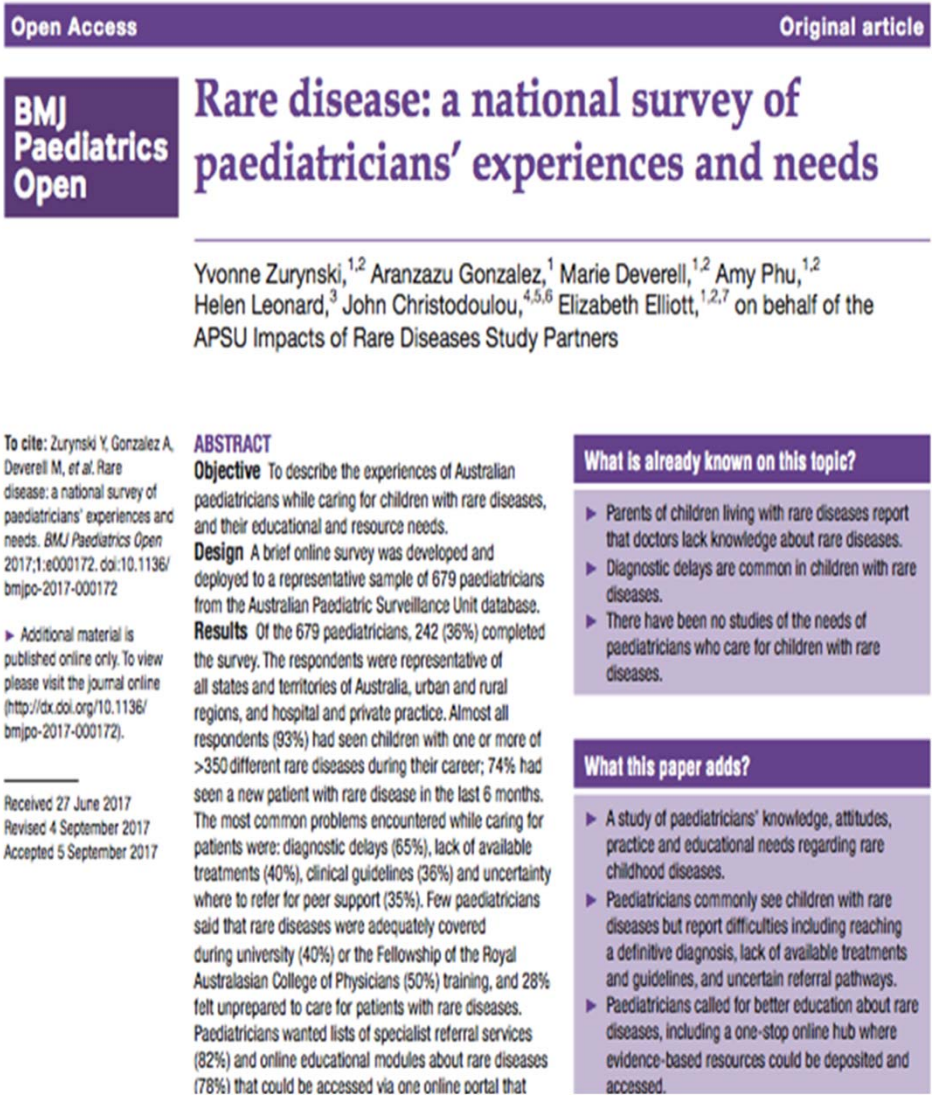
Paediatrician survey, representative (n=242)

- Inadequate training University (45%), FRACP (50%)
- Inability to make a diagnosis
  - Lack Dx guidelines/tests
  - Lack management guidelines/treatments
  - Difficulty accessing specialists
- 96% used internet resources to guide patient management
- 75% used smartphones, tablets in practice (F>M)
- 89% used APSU protocols, website, report
- ~30% aware of specific RD resources
  - Orphanet, NORD, EURODIS, AGSA

"I'll give it to you straight — This disease is almost *impossible* to pronounce."

Paediatricians want resources?

- 95% - printable resources for families
- 93% - RD support groups
- 82% - specialist services
- 78% - online educational
- 72% - smart phone/tablet apps on RD
- 70% - how to use online resources e.g. Orphanet.



Open Access Original article

BMJ Paediatrics Open

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

To cite: Zurynski Y, Gonzalez A, Deverell M, *et al*. Rare disease: a national survey of paediatricians' experiences and needs. *BMJ Paediatrics Open* 2017;1:e000172. doi:10.1136/bmjpo-2017-000172

► Additional material is published online only. To view please visit the journal online (http://dx.doi.org/10.1136/bmjpo-2017-000172).

Received 27 June 2017
Revised 4 September 2017
Accepted 5 September 2017

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

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.

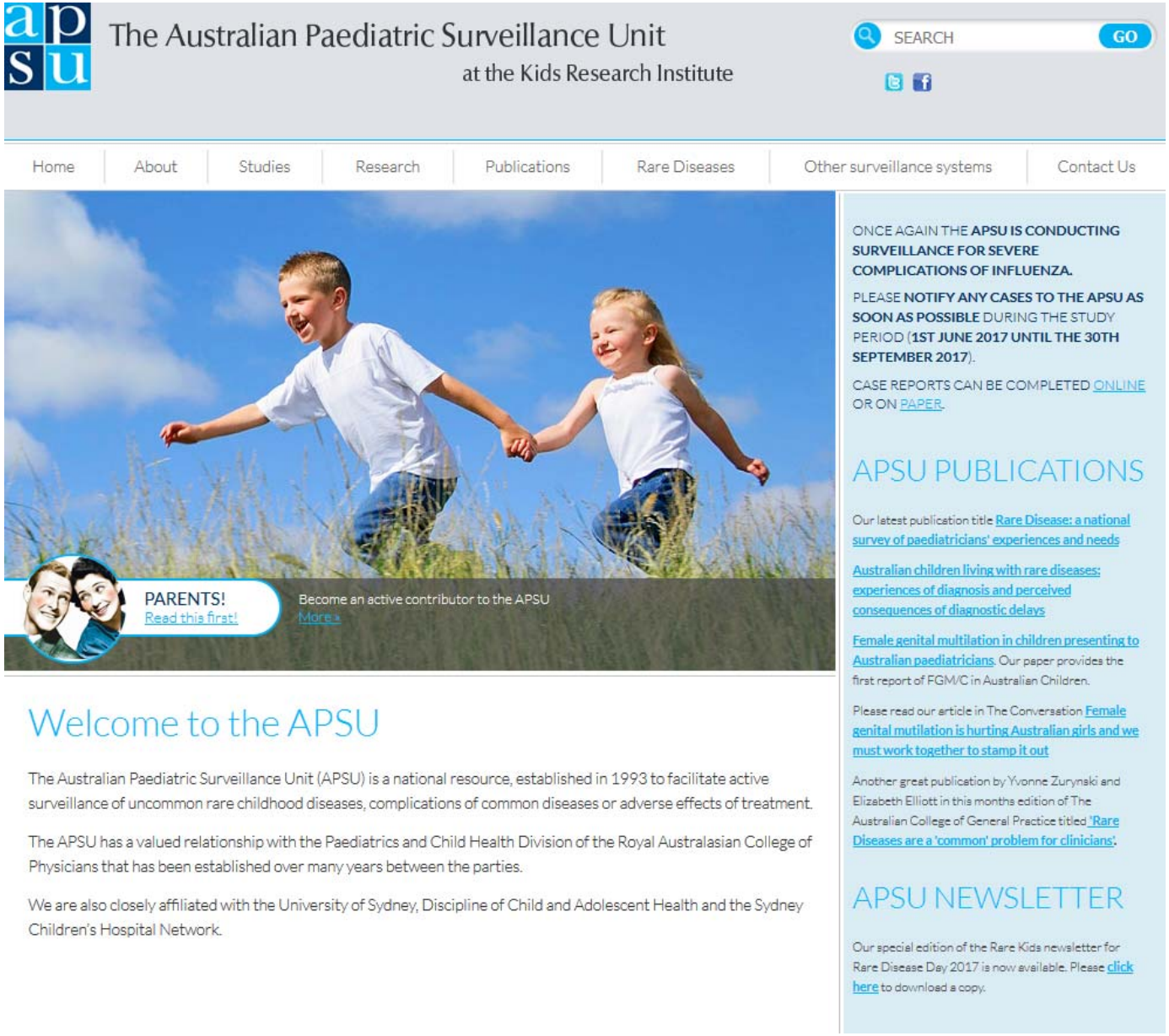
What this paper adds?

- 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.

Single portal linking all existing resources

EDUCATION & ADVOCACY

Education resources: paediatricians, parents



Potential to develop a “one-stop-shop” portal of resources

Education, advocacy: Female Genital Mutilation: its everyone's business. AHRC.

J Paediatr Child Health. 2010 Jan;46(1-2):2-4. doi: 10.1111/j.1440-1754.2009.01608.x. Epub 2009 Nov 23.

Call for a national plan for rare diseases.

Jaffe A, Zurynski Y, Beville L, Elliott E.
Respiratory Department, Sydney Children's Hospital, Randwick, Australia.

JPCH, 2009 "Australia requires a national plan.."

Abstract

Australia requires a national plan, similar to plans developed internationally, to address the impacts of rare diseases on individuals, the community and health services. Rare diseases often present in childhood, many are chronic, some life threatening and others associated with significant disability. However, diagnosis is often delayed, because of lack of knowledge and experience of health professionals and uncertainty about where to refer. Specialised health services are frequently lacking and specific therapies are often not available, partly because of lack of research funding directed towards rare diseases. A national plan would facilitate a coordinated response to service development, carer support, and health professional and community education, and would promote research and advocacy for affected children and their families.

Lancet, 2012:
"Without a well-considered national plan for RD, people will continue to receive sub-optimal care and support"

Australia makes up for lost time on rare diseases

Rare diseases collectively affect up to 10% of Australians, and advocates are lobbying national and state governments to accelerate efforts to develop a national plan. Tony Kirby reports.

Australia has some of the best health indicators of any high-income country, including increasing longevity bettered only by Japan. Yet in the sphere of rare diseases, the country has been lagging behind other developed nations. Australia has neither a coordinated national plan for rare diseases, nor a funded national umbrella organisation for these conditions, estimated collectively to affect some 6–10% of the population, equivalent to a total of 1·2 million Australians—the same number that have diabetes. There are some 400 000 children with rare diseases in Australia.

Although individual rare diseases, by definition, have a low prevalence, there are thousands

come up with a national plan for addressing rare diseases by 2013. America's National Institutes of Health (NIH) has a dedicated Office of Rare Diseases Research. Indeed, NIH director Frances Collins has made rare diseases one of the priorities of his leadership. Yet Australia is yet to adopt a national plan, despite the best efforts of advocates that include some of Australia's most well known public health and paediatrics experts.

"'...without a well-considered national plan for rare diseases... people with these conditions will continue to...receive sub-optimal care and support.'"

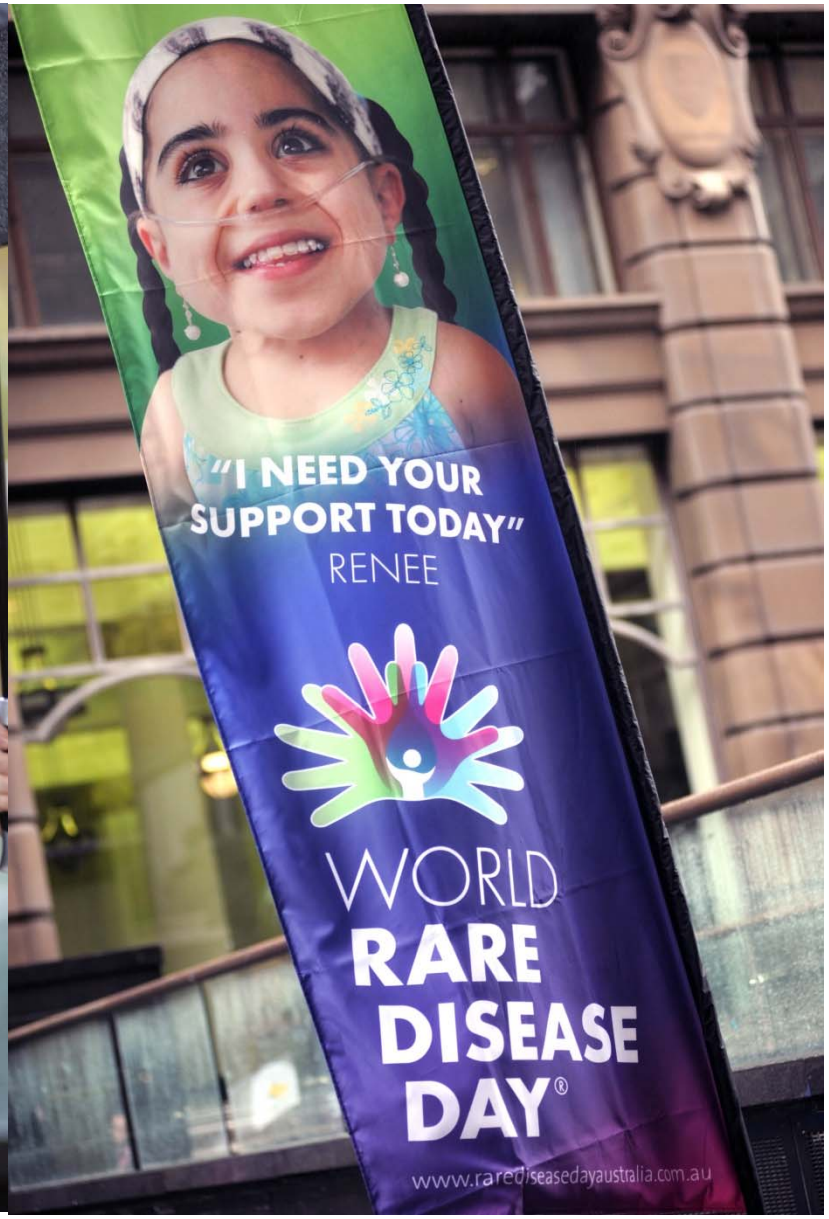
between 1 and 15 years caused by these conditions. However, obtaining a definitive diagnosis is often difficult and delayed. Neurological and intellectual disabilities occur in about half of all cases regardless of disease type and lead to loss of independence and opportunities. Families of those affected experience isolation, psychological and financial stress, and health professionals say they have inadequate access to information, education, and resources. Many children with rare diseases require multidisciplinary care and some have their education disrupted, while others have to abandon school altogether.

Elliott has, for two decades, run the Australian Paediatric Surveillance

Rare Disease Day

28 February 2010

www.rarediseaseday.org



Rare Disease Day 2012

The Australian Paediatric Surveillance Unit

National Polio Expert Committee

Western Pacific Polio accreditation committee

National FASD Technical Advisory Committee

Intergovernmental Committee on Drugs

National Rare Diseases Co-ordinating committee

Orphanet

SWF Medical Advisory Committee

SMILE Board

RARE Voices

Cure Kids Australia Board

RACP (DPCH)Research Committee

NHMRC, RACP, AMA, WHO committees (guidelines, position statement)

Specialist Clinical Committee MBS Review

Life saving drugs committee

At the stroke of a pen, a lifeline for Aviana

Johnny and Bethan McElwee with children Aviana, who has spinal muscular atrophy, and Hugo. Photo: Brook Mitchell

Until now, a drug that can save the lives of children with spinal muscular atrophy cost $375,000 a year. As of tomorrow, it will be $39.50 a script. NEWS PAGE 2

May 2018: $240 million Spinraza, Spinal Muscular Atrophy

The future?

- Funding
  - $500 mill genomics May budget
  - ? Clinical services, ? surveillance
- Maintain relevance
- Responsive to government needs
- ‘Niche’ role, avoid
- Deep phenotyping
  - assist interpretation of genomic data
- Use data for advocacy
  - National rare Disease Plan, novel services, support for families

THANKS

- Paediatricians who report to APSU
- RACP (Australian College of Paediatrics)
- Investigators
- Collaborating organisations
- Funders
- Scientific Review Panel
- Staff: Yvonne Zurynski, Katrina Williams, Anne Morris, Donna Rose, Greta Ridley, David Lester-Smith, Dannielle Handel, Ingrid Charters, Suzy Teutch, Gabrielle Williams, Susan Mildenhall, Amy Phu, Diane Redmond, Neil Heime, Marie Deverall, Jenny Peat, INOPSU, PAEDS, students

apsu.org.au