

APSU Summary Progress Report 2009

APSU highlights 2009

1. Rare Diseases Day – February 28, 2009

The APSU joined the global effort to raise awareness on rare childhood diseases by participating in Rare Diseases Day coordinated by the National Organisation of Rare Disorders (NORD) and Eurordis.

2. National Plan for Rare Diseases

On February 2 2009, APSU convened a group of researchers, clinicians and parent support groups to draft a coordinated national plan for rare diseases. The APSU was awarded a complimentary grant from the Australian Research Alliance for Children and Youth to progress the national plan. A copy of the draft national plan can be found at www.apsu.org.au.

3. Rare Diseases New Partnerships

New partnerships were forged with the Steve Waugh Foundation and SMILE Foundation which support families affected by rare diseases. These organisations will work together to host a workshop on Rare Diseases in February 2010 in Sydney and push forward a coordinated plan for rare diseases.

4. Royal Australasian College of Physicians: Physicians Week 17-21 May 2009

The APSU presented a session entitled "Paediatricians Dealing with Rare Diseases" at the Royal Australasian College of Physicians Week on Monday May 18 in Sydney. The program is available at www.physiciansweek.com/pch_program.asp.

5. Creswick Foundation Fellowship

As part of Dr Yvonne Zurynski's Creswick Fellowship, planned visits to various patient groups for people affected by rare diseases were conducted such as **Contact-a-Family; INVOLVE; Rare Diseases UK, EURORDIS and Orphanet**. Children and families affected by rare diseases and clinicians who care for these children need information about the condition, access to parent support groups, and medical and social services. Paediatric Surveillance Units are well placed to improve the availability of information on rare diseases that is accessible to patients, clinicians and the public. APSU will endeavour to provide patient accessible summaries for each condition studied in the future, including a summary of the condition, support groups if these exist, and any educational opportunities for parents. The British Paediatric Surveillance Unit has already begun this process. In addition, Orphanet has developed an educational program on rare diseases that is included in the medical curriculum in France to raise awareness of rare diseases among future clinicians, and to inform them of databases and search tools they may need when faced with diagnosing and managing people with rare diseases.

Surveillance studies

Commencing or concluding in 2009:

A study on *systemic lupus erythematosus (SLE)* commenced in October 2009 led by Dr Fiona Mackie. The study's objectives are to estimate the incidence, geographical distribution and ethnic background of SLE in Australian children aged ≤ 15 years, to describe the mode and severity of clinical presentations as well as one year outcome of SLE. In addition the clinical speciality of the initial treating physician as well as types of medications and any medication related adverse outcomes used in the management of paediatric SLE in the first year after diagnosis will be described.

Concluding:

- Neonatal herpes simplex virus concluded in December 2009 and 555 cases were reported.
- Neuromuscular disorders of childhood concluded in December 2009 and 373 cases were reported. Data is currently being analysed by investigators.

Contributors to the APSU

Contributors to the APSU are clinicians working in paediatrics and child health in Australia. Most (about 80%) are general paediatricians with or without a special interest. In addition 6% are neonatologists, 3.5% are surgeons, 2.6% are geneticists and 2% are emergency physicians.

Summary of Surveillance Data

In 2009, 1330 clinicians participated in the monthly surveillance of 15 conditions, with an overall response rate of 90%. The majority of clinicians (82%) reported by e-mail. Detailed data were obtained on 85% of all reported cases.

Respondent Workload

The workload in completing questionnaires is low. During 2009 the majority of clinicians, (79.4%) had no cases to report, 12.9% reported one case, 3.6% reported two cases and 4.1% reported three or more cases.

The table shows the reported rates of disease for conditions studied through the APSU in 2009. For conditions where cases were ascertained through additional complementary sources e.g. mandatory reporting systems and laboratory surveillance (including perinatal exposure to HIV, acute flaccid paralysis and Rett syndrome), cases from more than once source have been included to estimate the rate of disease.

Reported rate for each condition studied to December 2009

Conditions Under Surveillance	Study period	Duration of study (years)	Total notifications	Questionnaires returned, n (%)	Total confirmed cases for duration of study*	Reported Rate for duration of study (per 10 ⁵ per annum)
Infectious/vaccine preventable conditions						
Acute flaccid paralysis	Mar 1995 ongoing	15	876	806 (92)	559	0.93 ^b
Acute rheumatic fever	Oct 2007 ongoing	2.5	180	159 (88)	104	1.00 ^b
Congenital cytomegalovirus (cCMV) infection	Jan 1999 ongoing	11	397	295 (74)	161	5.53 ^a
Neonatal herpes simplex virus infection	Jan 1997 ongoing	13	233	223 (96)	117	3.43 ^a
Perinatal exposure to HIV infection	May 1993 ongoing	16.5			362	8.40 ^a
Perinatal HIV infection					65	1.51 ^a
Serious complications of seasonal influenza	Jul 2009 – Sep 2009	0.25	146	142 (97)	101	†
Intussusception	May 2007 ongoing	2.5	287	231 (80)	154	10.75 ^c
Congenital rubella (with defects)	May 1993 ongoing	16.5	110	106 (96)	51	0.07 ^d
Congenital Varicella	May 2006 ongoing	3.5	12	12 (100)	2	0.20 ^a
Neonatal Varicella	May 2006 ongoing	3.5	26	23 (88)	17	1.67 ^a
Severe complications of Varicella infection	May 2006 ongoing	3.5	58	47 (81)	37	0.26 ^b
Congenital/genetic disorders						
Neuromuscular disorders	Jan 2007 ongoing	3	387	304 (79)	207	1.57 ^d
Rett syndrome	Jan 2000 ongoing	10	284	276 (97)	155	0.74 [†]
Other injury/illness						
Vitamin K deficiency bleeding	May 1993 ongoing	16.5	142	139 (98)	33	0.77 ^a
Systemic lupus erythematosus	Oct 2009 ongoing	1	7	5(71%)	3	†

* Total confirmed cases according to case definition in the study protocol

a. Reported incidence per 100,000 live births

b. Reported incidence per 100,000 children < 15 years

c. Reported incidence per 100,000 children ≤ 24 months

d. Reported incidence per 100,000 children < 16 years

f. Reported incidence per 100,000 females < 16 years

† Due to the limited surveillance period a reported rate cannot be calculated

APSU Website

As the unit expands its activities, the website continues to grow. The number of hits in 2009 was 11,798 and is a clear indication that the site is accessed by a widely.

Publications/Abstracts and Presentations in 2009

1. Benitez-Aguirre PZ, Wood NJ, Biesheuvel C, Moreira C, Munns CF. The natural history of vitamin D deficiency in African refugees living in Sydney. *Medical Journal of Australia* 2009; 190(8): 426-428.
2. Blyth C, Best E, Jones C, Nourse C, Goldwater P, Daley A, Burgner D, Henry G, Palasanthiran P. Nontuberculous mycobacterial infection in children: a prospective national study. *Pediatric Infectious Diseases Journal* 2009; 28(9):801-805.
3. Grenier D, Lynn R, Zurynski Y on behalf of all national paediatric surveillance unit investigators. Public health impacts of paediatric surveillance units. *Paediatrics and Child Health* 2009; 45:499-504.
4. He S, Zurynski Y, Elliott E. Evaluation of a national resource to identify and study rare diseases: the Australian Paediatric Surveillance Unit. *Journal of Paediatrics and Child Health* 2009; 45(9): 498-504.
5. Lester-Smith D, Zurynski Y, Booy R, Festa MS, Kesson AM, Elliott EJ. The burden of childhood influenza in a tertiary paediatric setting. *Communicable Diseases Intelligence* 2009; 33(2): 208-214.
6. Madden S, Morris A, Zurynski Y, Kohn M, Elliott E. The burden of eating disorders in children aged 5-13 years in Australia. *Medical Journal of Australia* 2009; 8:410-414.
7. McDonald A, Zurynski Y, Wand HC, Giles M, Elliott E, Ziegler J, Kaldor J. Perinatal exposure to HIV among children born in Australia. *Medical Journal of Australia* 2009; 190:416-420.
8. Stewardson AJ, Roberts JA, Beckett CL, Prime HT, Loh PS, Thorley BR, Daffy JR. Imported Case of Poliomyelitis, Melbourne, Australia, 2007. *Emerging Infectious Diseases* 2009; www.cdc.gov/eid. Vol.15, No.1.

Abstracts

1. Lester-Smith D, Zurynski Y, Festa M, Kesson A, Booy R, Elliott E. Seasonal surveillance for serious complications of influenza in children using the Australian Paediatric Surveillance Unit. *Journal of Paediatrics and Child Health* 2009; 45(11):A4.
2. McDonald A, Zurynski Y, Nadew K, Elliott E, Kaldor J, Ziegler J. Prevention of mother-to-child HIV transmission in Australia, 1982-2005. *Journal of Paediatrics and Child Health* 2009; 43(7-8):A11.
3. Smith MT, Lester-Smith D, Zurynski Y, Noonan S, Sholler G, Carapetis JR, Elliott E. Acute Rheumatic Fever Presentations to a Tertiary Hospital. *Journal of Paediatrics and Child Health* 2009; 45(11):A8
4. Wood N, Royle J, Zurynski Y, Adams J, Booy R, Buttery J, Elia S, Gold M, Heath C, McKay N, Marshall H, McIntyre P, Phillips A, Pym M, Rhind L, Richmond P, Wall K, Elliott E. Seizures in Infants: results from Paediatric Active Enhanced Disease Surveillance (PAEDS). *Journal of Paediatrics and Child Health* 2009; 45(11):A22.
5. Zurynski Y, He S, Elliott E. What do paediatricians think of the Australian Paediatric Surveillance Unit? *Journal of Paediatrics and Child Health* 2009; 45(9): A10.
6. Zurynski Y, Royle J, Booy R, Elliott E, Buttery J, Marshall H, Gold M, Richmond P, McIntyre P, Wood N, Rhind L, Pym M, McKay N, Heath C. A New Surveillance Mechanism for Childhood Conditions: Paediatric Active Enhanced Disease Surveillance (PAEDS). *Journal of Paediatrics and Child Health* 2009; 45(9): A22.

Presentations

1. Elliott E. Fetal Alcohol Spectrum Disorders (FASD) in Australia: A Monograph. Presentation to the Inter-Governmental Committee on Drugs (IGCD) on behalf of the IGCD FASD Working Party. Perth, February 2009
2. Elliott E. Alcohol in pregnancy. 16th Annual Women's Health Conference. Queensland, February 2009. (Keynote)
3. Elliott E, Zurynski Y. Australian Paediatric Surveillance Unit (APSU) and Paediatric Active Enhanced Disease Surveillance (PAEDS): Latest developments in paediatric surveillance. National Centre for Immunisation Research Surveillance (NCIRS) Journal Club. Sydney, March 2009.
4. Elliott E. Pre-Pregnancy Care - Can we do better? Women's Hospitals Australasia Scientific Meeting. Parkville, May 2009
5. Elliott E. A national plan for rare diseases. APSU Session. RACP Physicians Week. Sydney, May 2009.

6. Jaffe A. Australian rare lung diseases registry: how disease registries can inform your practice. RACP Physicians Week. Sydney, May 2009.
7. Mackie F. Systemic lupus erythematosus. Proposed new conditions for APSU surveillance. RACP Physicians Week. Sydney, May 2009.
8. Payne J, France K, Henley N, D'Antoine H, Bartu A, O'Leary C, Elliott E, Bower C & the Alcohol and pregnancy Steering Committee. Bridging the evidence-practice gap. PHAA 39th Annual Conference. Canberra, September 2009.
9. Zurynski Y. Subdural haematomas in children <2years. Proposed new conditions for APSU surveillance. APSU Session. RACP Physicians Week. Sydney, May 2009.

Thank you to every clinician who participates in APSU surveillance, for your continued support of this important work. We look forward to your continued participation.