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.

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 specialty 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

<table>
<thead>
<tr>
<th>Conditions Under Surveillance</th>
<th>Study period</th>
<th>Duration of study (years)</th>
<th>Total notifications</th>
<th>Questionnaires returned, n (%)</th>
<th>Total confirmed cases for duration of study*</th>
<th>Reported Rate for duration of study (per 10^5 per annum)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Infectious/vaccine preventable conditions</strong></td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Acute flaccid paralysis</td>
<td>Mar 1995 ongoing</td>
<td>15</td>
<td>876</td>
<td>806 (92)</td>
<td>559</td>
<td>0.93^a</td>
</tr>
<tr>
<td>Acute rheumatic fever</td>
<td>Oct 2007 ongoing</td>
<td>2.5</td>
<td>180</td>
<td>159 (88)</td>
<td>104</td>
<td>1.00^b</td>
</tr>
<tr>
<td>Congenital cytomegalovirus (cCMV) infection</td>
<td>Jan 1999 ongoing</td>
<td>11</td>
<td>397</td>
<td>295 (74)</td>
<td>161</td>
<td>5.53^c</td>
</tr>
<tr>
<td>Neonatal herpes simplex virus infection</td>
<td>Jan 1997 ongoing</td>
<td>13</td>
<td>233</td>
<td>223 (96)</td>
<td>117</td>
<td>3.43^d</td>
</tr>
<tr>
<td>Perinatal exposure to HIV infection</td>
<td>May 1993 ongoing</td>
<td>16.5</td>
<td></td>
<td></td>
<td>762</td>
<td>8.40^e</td>
</tr>
<tr>
<td>Severe complications of seasonal influenza</td>
<td>Jul 2009 – Sep 2009</td>
<td>0.25</td>
<td>146</td>
<td>142 (97)</td>
<td>101</td>
<td>†</td>
</tr>
<tr>
<td>Intussusception</td>
<td>May 2007 ongoing</td>
<td>2.5</td>
<td>287</td>
<td>231 (80)</td>
<td>154</td>
<td>10.75^e</td>
</tr>
<tr>
<td>Congenital rubella (with defects)</td>
<td>May 1993 ongoing</td>
<td>16.5</td>
<td>110</td>
<td>106 (96)</td>
<td>51</td>
<td>0.07^f</td>
</tr>
<tr>
<td>Congenital Varicella</td>
<td>May 2006 ongoing</td>
<td>3.5</td>
<td>12</td>
<td>12 (100)</td>
<td>2</td>
<td>0.20^g</td>
</tr>
<tr>
<td>Neonatal Varicella</td>
<td>May 2006 ongoing</td>
<td>3.5</td>
<td>26</td>
<td>23 (88)</td>
<td>17</td>
<td>1.67^a</td>
</tr>
<tr>
<td>Severe complications of Varicella infection</td>
<td>May 2006 ongoing</td>
<td>3.5</td>
<td>58</td>
<td>47 (81)</td>
<td>37</td>
<td>0.26^h</td>
</tr>
<tr>
<td><strong>Congenital/genetic disorders</strong></td>
<td></td>
<td></td>
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<tr>
<td>Neuromuscular disorders</td>
<td>Jan 2007 ongoing</td>
<td>3</td>
<td>387</td>
<td>304 (79)</td>
<td>207</td>
<td>1.57^g</td>
</tr>
<tr>
<td>Rett syndrome</td>
<td>Jan 2000 ongoing</td>
<td>10</td>
<td>284</td>
<td>276 (97)</td>
<td>155</td>
<td>0.74^i</td>
</tr>
<tr>
<td><strong>Other injury/illness</strong></td>
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<td></td>
<td></td>
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<tr>
<td>Vitamin K deficiency bleeding</td>
<td>May 1993 ongoing</td>
<td>16.5</td>
<td>142</td>
<td>139 (98)</td>
<td>33</td>
<td>0.77^a</td>
</tr>
<tr>
<td>Systemic lupus erythematosus</td>
<td>Oct 2009 ongoing</td>
<td>1</td>
<td>7</td>
<td>5(71%)</td>
<td>3</td>
<td>†</td>
</tr>
</tbody>
</table>

* 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


Abstracts


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)


4. Elliott E. Pre-Pregnancy Care - Can we do better? Women's Hospitals Australasia Scientific Meeting. Parkville, 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.