

# rare

# Kids



Australian Paediatric Surveillance Unit **NEWSLETTER** March 2012



## New Studies for 2012

Teams of researchers have been working hard to develop two new protocols which have been added to the APSU report card in January 2012.

**Food Protein Induced Enterocolitis Syndrome (FPIES)**, is a paediatric non-immunoglobulin E mediated allergic disorder triggered by ingestion of certain food protein(s) and presenting mostly in children aged < 12 months. Typically children present 2–4 hours after ingestion of a food protein recently introduced into the diet with profuse vomiting and some subsequently develop diarrhoea. Some may present in a moribund state with pallor, floppiness, reduced body temperature, hypovolemic shock and/or metabolic acidosis. The prognosis for FPIES is good and most attain tolerance to the food trigger by 3 years of age.

The APSU study will estimate the incidence of FPIES in Australia and provide important new knowledge to raise awareness of this rare allergic condition and to support development of educational material for clinicians and parents, and formulation of clinical practice guidelines. *To find out more about FPIES please contact Study Chief Investigator: Dr Sam Mehr [sam.mehr@health.nsw.gov.au](mailto:sam.mehr@health.nsw.gov.au)*

### **Sudden Unexpected Early Neonatal Death Or Collapse In Previously Healthy Term Infants In The First 7 Days Of Life.**

The incidence of sudden unexpected and unexplained death or neonatal collapse is reported as between 0.035/1000 to 0.4/1000 live births. Although rare, greater than half of these infants die and the majority of survivors have significant long term neurodevelopmental morbidities. There is currently no national system available in Australia for investigating and reporting these cases.

We aim to establish the current incidence of sudden unexplained death or collapse in the early neonatal period (first 7 days of life) in Australia. In the reported literature many of these infants are found face down on the mother's breasts suggesting that airway compromise may be a contributing factor. Other risk factors include maternal analgesia, bed-sharing and prone sleeping. We aim to document the risk factors and outcomes for such cases in Australia.

Information obtained through the APSU study will lead to the development and implementation of policy on safe postnatal management of healthy newborns and development of educational materials for parents, carers and health professionals.

To find out more about the Sudden Neonatal Death study please contact Prof Heather Jeffery [heather.jeffery@sydney.edu.au](mailto:heather.jeffery@sydney.edu.au)

## Welcome and Thank You

Welcome to a new look newsletter for the Australian Paediatric Surveillance Unit. We believe the new title "Rare Kids" is very much in keeping with our focus on research and surveillance to support better outcomes for children who have rare conditions.

We aim to provide you with news of APSU activities and achievements 3 times per year: in April, August and December.

We would be pleased to receive any suggestions to improve the newsletter, and we hope that you will enjoy this inaugural edition.

This newsletter will be distributed via e-mail and will also appear on the APSU website [www.apsu.org.au](http://www.apsu.org.au)

As we commence another successful year, we would like to take a moment to thank the 1380 paediatricians and other child health specialists who actively participate in APSU surveillance. We very much appreciate the time and effort taken by each of you to return the report cards each month and to provide information on cases. We look forward to your continuing support in 2012.

Thanks also go to our funders and supporters who are listed on page 4 of this newsletter. Without your support the important work of the APSU could not continue.

APSU Team

**For more information on new and existing APSU studies please visit our website [www.apsu.org.au](http://www.apsu.org.au).**

**Protocols and Questionnaires for all past and current studies are available for download from here, simply click on the "studies" tab on the home page.**

## New Studies introduced in 2011

We introduced 2 new conditions to APSU in 2011: Juvenile onset Recurrent Respiratory Papillomatosis (JoRRP) led by Dr Daniel Novakovic and Cryopyrin Associated Periodic Syndromes (CAPS) led by Dr Sam Mehr.

CAPS is an extremely rare condition and not suitable for continued surveillance. We therefore used a once-off report card to provide a snap-shot of the number of cases that have been diagnosed in Australia in 2011. Thank you to all clinicians reporting cases of CAPS.

We also continued surveillance for children hospitalised with severe complications of influenza during the months July to September. This is the 4<sup>th</sup> year of severe influenza surveillance by the APSU and provides unique data from before, during and after the 2009 H1N1 influenza pandemic.

## Our Website: [www.apsu.org.au](http://www.apsu.org.au)

The APSU website is updated regularly and is full of useful information about the conditions being studied, reports, news on APSU activities and lists of publications. All study materials are available for download under the "Current Studies" tab on the website.

Did you know that APSU also provides *Information Sheets for Parents*, carers and the community? For many of the new conditions added to the APSU report card, investigators have developed information sheets which include a description of the condition and diagnosis, the rationale for doing a surveillance study and links to resources that may be useful for parent or carers, e.g. parent support groups or links to relevant foundations, government and non-government organisations. The information sheets can be downloaded and printed by parents or health professionals. We hope you will find this a useful resource. Many thanks to the Royal Australasian College of Physicians for hosting the APSU website on their server.

## APSU Biennial Research Report 2009-2010

The APSU Biennial Research Report for 2009-2010 which highlights major development and achievements will be on the APSU website before the end of February. The report provides summary surveillance data including incidence estimates and key findings for each of the conditions studied. We are very grateful to the expert teams of clinicians and researchers working on each of the surveillance studies for their efforts to make this possible. The report also provides information about upcoming studies, including rationale and case definitions.

To acknowledge the efforts of all clinicians who report to the APSU we print lists of all who return all 12 report cards during the year and those who report cases. The work of the APSU is not possible without your continued participation – THANK YOU!

**Wine Prize!** Once again Mount Majura Winery supported the annual wine prize. Every year we choose at random 12 clinicians who have returned all 12 of their report cards to the APSU and 12 clinicians who reported cases and provided completed questionnaires. All prize winners should have received their bottle of wine by now – we hope you will enjoy!

If you would like a hard-copy of the report please contact the APSU on (02) 9845 3005 or email us at [apsu@chw.edu.au](mailto:apsu@chw.edu.au)

## APSU Online Reporting System

The number of clinicians reporting via e-mail increased significantly from 68% in 2008 to 83% in 2011. *We now offer web-based reporting in NSW – an innovation which saves approximately 20 hours of data entry each month!*

In late 2010 APSU received support from the Information and Communication Technology Department of the University of Sydney under their e-Research platform to develop a web-based reporting system. This will make reporting more efficient for clinicians and for APSU staff who currently have to manually enter 1380 responses into the APSU database each month. The web-based reporting system was trialed in NSW in January 2011 and comprised 200 NSW contributors. At present all NSW clinicians who previously responded by e-mail are now reporting via the web-based system.

We plan to implement a staged roll-out to other states over 2012 and we hope you will embrace this efficient way of reporting.

If you practice in NSW you are now receiving an e-mail with a weblink which enables you to record your response directly.

**Remember!** Click this link **nothing to report or case to report** which appear in your monthly e-mail card – don't click reply! Please refer to screen shots on the following page.

Information regarding the new reporting system can be accessed and downloaded via our website [www.apsu.org.au](http://www.apsu.org.au) – click on the link provided on home page under "APSU Online Reporting System". Alternatively you can contact us on 02 9845 3005.

## RACP Congress 2012 - Brisbane

The 2012 RACP Future Directions in Health Congress will be held at the Brisbane Convention and Exhibition Centre on 6<sup>th</sup> – 9<sup>th</sup> May. The theme for the 2012 Congress is *Disease and Injury Prevention*. The 4 day event remains the premier annual event on the RACP calendar.

The APSU will once again host a session in keeping with the theme of the Congress entitled: *The significance of injury surveillance data in Australia and internationally* and we would like to extend a warm invitation to attend the Congress and the APSU session. To view the draft program or for more information about the Congress please visit <http://www.racpcongress2012.com.au/>

## Rare Diseases Educational Workshop 7<sup>th</sup> March 2012

To mark Rare Disease Day, APSU will once again host a workshop for clinicians and interested families. The workshop will feature presentations on the latest developments in rare diseases research, policy and advocacy in Australia, while providing an opportunity for clinicians and families alike to come away with practical information about health services and family support organisations. Importantly, we have invited families living with rare disease to provide insight into their experiences.

The workshop will be held at the Kids Research Institute, the Children's Hospital at Westmead in Sydney from 9am to 5pm on 7<sup>th</sup> March. Further information available soon on the APSU website.

## Rett Syndrome information brochure and Scoliosis guidelines

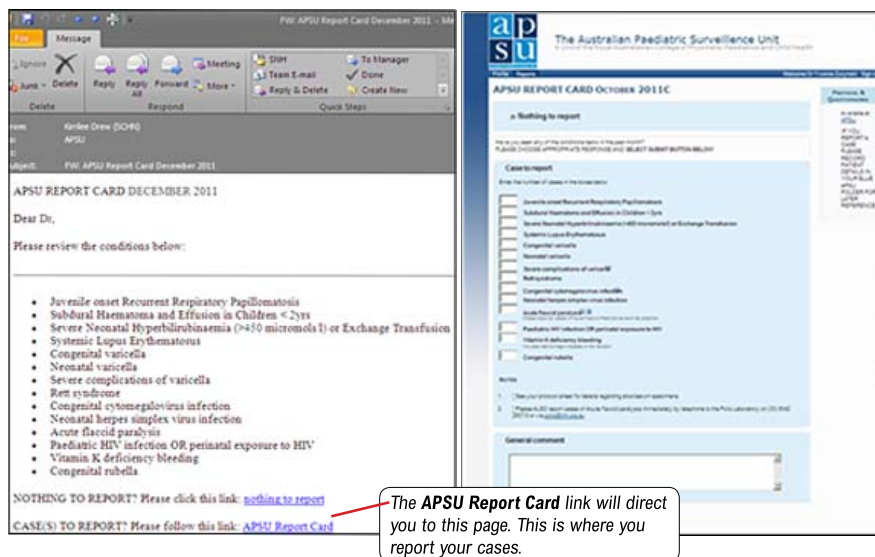
The Telethon Institute for Child Health Research has recently released a new RETT Syndrome Brochure and Scoliosis Guideline Booklet. The brochure highlights the achievements of the Australian Rett Syndrome study since its establishment in 1993. It also details the inner working of the unit and reinforces the overall aim of the unit. A copy of this brochure is enclosed for your reference.

The Scoliosis Guideline Booklet provides an outline of recommended management guidelines for scoliosis in Rett Syndrome and lays the groundwork for parents and clinicians alike by providing key background information. The booklet also contains a selection of firsthand accounts from parents who have a child with scoliosis.

If you would like any further information on either of the documents please contact 08 9489 7789 or [aussierett@icmr.uwa.edu.au](mailto:aussierett@icmr.uwa.edu.au)

**Did You Know??**  
**One in 10 Australians will be affected with a rare disease in their lifetime!**





## Rare Disease Day 2012

In 2011, 56 countries around the world participated in raising awareness of the impacts of rare diseases through Rare Disease Day activities and Rare Disease Day 2012 is shaping up to be bigger and better still <http://www.rarediseaseday.org/tp://www.rarediseaseday.org/>

To mark Rare Disease Day 2012, APSU will once again join family support organisations including AGSA, Steve Waugh Foundation and SMILE Foundation in awareness raising activities at Martin Place in the Sydney CBD on 29<sup>th</sup> February. Please keep a close watch on the APSU website for further information. The theme for Rare Disease Day 2012 is *"SOLIDARITY: Rare but Strong Together"*.

APSU will also hold an Educational Workshop on Rare Diseases on 7<sup>th</sup> March at the Kids Research Institute, Westmead, Sydney. For details please see [www.apsu.org.au](http://www.apsu.org.au) or call APSU on 9845 3005



Photo courtesy of Steve Waugh Foundation photographer Jack Atley

Rare Disease Day Function 2011 hosted by the Steve Waugh Foundation, Sydney

## Perspectives on Establishing a Coordinated Approach to a National Plan - Fremantle, 17-20 April

An International Rare Diseases Symposium was held in Fremantle, WA on 17-20 April and convened by the Office of Population Health Genomics, Public Health Division, Department of Health, Western Australia and the Australian Paediatric Surveillance Unit (APSU).

The National Organising Committee for this symposium included Elizabeth Elliott and Yvonne Zuryski from the APSU, and Kathryn North from the Neurogenetics Research Unit and the Institute for Neuromuscular Research, The Children's Hospital at Westmead. The conference was attended by over 300 national and international delegates including patients, representatives of support groups and charitable organisations, researchers, clinicians, government officials and representatives from industry. The program included Australian and international speakers, while including opportunities for people affected by a rare disease to present their perspective. The discussion forums resulted in a number of recommendations including the establishment of an umbrella organisation for rare diseases in Australia and the progression of the Rare Diseases National Plan.

For more information please visit: [www.raredisease.com.au](http://www.raredisease.com.au).

## APSU and PAEDS Selected Key Publications for 2011

- Buttery JP, Danchin MH, Lee KJ, Carlin JB, McIntyre PB, Elliott EJ, Booy R, Bines JE, for the PAEDS/APSU Study Group. Intussusception following rotavirus vaccine administration: Post-marketing surveillance in the National Immunization Program in Australia. *Vaccine* 2011; 29(16): 3061-3066.
- Fehr S, Bebbington A, Ellaway C, Rowe P, Leonard H, Downs J. Altered Attainment of Developmental Milestones Influences the Age of Diagnosis of Rett Syndrome. *Journal of Child Neurology* 2011; 26(8): 980-987.
- Fehr S, Bebbington A, Nassar N, Downs J, Ronen GM, De Klerk N, Leonard H. Trends in the Diagnosis of Rett Syndrome in Australia. *Pediatric Research* 2011; 70(3): 313-319.
- Hendrie D, Bebbington A, Bower C, Leonard H. Measuring use and cost of health sector and related care in a population of girls and young women with Rett syndrome. *Research in Autism Spectrum Disorders* 2011; 5(2): 901-909.
- Khandaker G, Booy R, Zuryski Y, Lester-Smith D, Kesson A, Heron L, Dwyer D. Clinical features, oseltamivir treatment and outcomes in infants aged less than 12 months with laboratory confirmed influenza A in 2009. *2011 Antiviral Therapy* 2011; DOI 10.3851/IMP1848.
- Khandaker G, Marshall H, Peadon E, Zuryski Y, Burgner D, Buttery J, Gold M, Nissen M, Elliott E, Burgess M, Booy R. Congenital and neonatal varicella: impact of the national varicella vaccination program in Australia. *Archives of Disease in Childhood* 2011; 96(5): 453-6.
- Lim A, Cranswick N, South M. Adverse events associated with the use of complementary and alternative medicine in children. *Archives of Diseases in Childhood* 2011; 96(3): 297-300.
- McMullan B, Palasanthiran P, Jones C, Hall B, Robertson P, Howard J, Rawlinson W. Congenital cytomegalovirus — time to diagnosis, management and clinical sequelae in Australia: opportunities for earlier identification. *Medical Journal Australia* 2011; 194(12): 625-629.
- Smith M, Lester-Smith D, Zuryski Y, Elliott E, Noonan S, Carapetis J. Persistence of acute rheumatic fever in a tertiary children's hospital. *Journal of Paediatrics and Child Health* 2011; 47(4):198-203.
- Smith M, Zuryski Y, Lester-Smith D, Elliott E, Carapetis J. Rheumatic fever - Identification, management and secondary prevention. *Aust Fam Physician* 2012, 41 (1-2):31-5
- Argent E, Emdor P, Monagle P, Mowat D, Petterson T, Russell S, Sachdev R, Stone C. and Ziegler, D. S. (2011), Australian Paediatric Surveillance Unit study of haemoglobinopathies in Australian children. *Journal of Paediatrics and Child Health*. doi: 10.1111/j.1440-1754.2011.02236.x
- Elliott E. J, Zuryski Y. A, Walls T, Whitehead B, Gilmour R, Booy, R. and Members of SWINet, Novel inpatient surveillance in tertiary paediatric hospitals in New South Wales illustrates impact of first-wave pandemic influenza A H1N1 (2009) and informs future health service planning. *Journal of Paediatrics and Child Health* 2011; doi: 10.1111/j.1440-1754.2011.02240.x

## APSU ARC Linkage Project Grant

Psychosocial and Economic Impacts of Rare Diseases on Australian Children, Families and Health Professionals

*Investigators:* A/Prof Yvonne Zurynski, Prof Elizabeth Elliott, A/Prof Helen Leonard, Prof John Christodoulou

*Partner Institutions:*

Royal Australasian College of Physicians  
The Children's Hospital at Westmead  
Steve Waugh Foundation  
SMILE Foundation  
The Association of Genetic Support Australasia

We lack systematically collected data to demonstrate the impacts of rare diseases in Australia. The APSU's ARC Linkage project provides an opportunity to address this knowledge gap by addressing the problem from 3 different angles:

- 1) A survey of families caring for a child with a rare disease will provide information on psychosocial, economic, educational and health and support service needs.
- 2) A survey of Australian paediatricians will highlight current awareness of rare diseases, needs for information about referral pathways and diagnostic and clinical services that can be accessed for their patients, needs for educational materials and opportunities, and the preferred way to deliver these.
- 3) An audit of admissions and presentations to the Children's Hospital at Westmead and specifically the Genetic Clinics in the hospital to demonstrate service usage and to estimate the costs of services, with the assistance of the Management Support and Analysis Unit and Medical Records Department at the hospital.



Delegates at the 7<sup>th</sup> INoPSU Conference, Montreux, Switzerland

## International Activities: 7<sup>th</sup> INoPSU Conference – Montreux 2011

In September this year, the Swiss Paediatric Surveillance Unit hosted the 7<sup>th</sup> INoPSU scientific meeting by Lake Lemman in Montreux. Representatives from Australia, Canada, UK, the Netherlands, Portugal and the newest member of INoPSU the Belgium Paediatric Surveillance Unit attended this important event.

The meeting provided the ideal forum for representatives from National Units around the globe to share and discuss surveillance methodology, key findings, publications, and potential for conducting simultaneous surveillance for some conditions internationally.

The scientific meeting brought together more than 80 conference delegates comprising of paediatricians, epidemiologists and health workers. The meeting showcased the amazing work undertaken by the Swiss, particularly their work on HUS, shaken baby syndrome and hyperbilirubinaemia.

Yvonne Zurynski presented on behalf Craig Munns and the Vitamin D Deficiency Rickets Study Group and compared Australian data with that obtained in Canada. Australian data showed that Vitamin D deficiency is not rare among the African refugee population who should be targeted for prevention and intervention. The important work of the APSU on engaging with parent support groups and involving families in research was also highlighted. Presentations, including this one can be viewed and downloaded from <http://www.INoPSU.com/publications/index.html>.

The 8<sup>th</sup> INoPSU Meeting will be held in conjunction with the International Paediatric Association conference in Melbourne from 24<sup>th</sup> -29<sup>th</sup> August 2013. The meeting will also be celebrating the INoPSU Meeting's 15<sup>th</sup> anniversary. For more information on this event please visit <http://www.ipa-world.org/>. 2013 is also the year that the APSU will be celebrating its 20 year anniversary.

## National Coordinated Plan

APSU has been advocating for the development of a national coordinated plan to address the many complex impacts of rare diseases on families, clinicians and the community. APSU produced and distributed a postcard to raise awareness of rare diseases and to encourage the community to support the development of a plan for Rare Diseases (please see the draft plan on [www.apsu.org.au](http://www.apsu.org.au)).

Please show your support for the development of a National Plan for Rare Diseases by participating in our brief on-line survey: [www.apsu.org.au](http://www.apsu.org.au)

## New email addresses for APSU Staff

Under the new Sydney Children's Hospital Network structure, staff emails have changed to firstname.surname@health.nsw.gov.au. This change is applicable for all existing APSU staff emails.

Old email addresses ([apsu@chw.edu.au](mailto:apsu@chw.edu.au)) will be routed to the new email address for at least the next 2 years however we ask that you make note of the change of email address and use these for future correspondence.

## The Staff at APSU

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