

rare

Kids



Australian Paediatric Surveillance Unit **NEWSLETTER**

JULY 2014

New Study—MECP2 Duplication syndrome

Individuals with two or more copies of the MECP2 gene share a distinct clinical phenotype and may present with a range of general indications such as developmental delay, intellectual disability, dysmorphic features, multiple congenital anomalies, autism, epilepsy, short stature, or failure to thrive. This study, led by Professor Helen Leonard, will improve our understanding of the core clinical features of MECP2 duplication syndrome and its epidemiology. MECP2 Duplication syndrome may be diagnosed by chromosome microarray testing which is now widely available. Please report all cases diagnosed from June 2014 according to the protocol and questionnaire distributed to you in May and available on the APSU website www.apsu.org.au

Studies concluding

Food Protein Induced Enterocolitis (FPIES)

This study concluded in April 2014. There were 340 notifications of FPIES between January 2012 and April 2014. The investigators group led by Dr Sam Mehr are currently analysing the data. Please report any additional cases and return questionnaires on all cases you have already reported. Thank you for reporting cases of FPIES.

Eosinophilic Oesophagitis (EoE)

This study concluded with the June 2014 report card. Please report any outstanding additional cases that you have seen since July 2013. If you have reported cases but have not yet provided a completed questionnaire for each case please do as soon as possible. On behalf of the Investigators Group led by Professor Dianne Campbell, thank you for reporting cases of EoE.

Severe complications of Varicella Infection

The number of severe hospitalised cases of varicella infection has reduced steadily since the introduction of the varicella vaccine onto the National Immunisation Program (NIP) in late 2005. This study concluded in December 2013 and the investigators, led by Professor Robert Booy are currently analysing data. Thank you for contributing cases to this study.

Change in Case definition

Juvenile onset Recurrent Respiratory Papillomatosis (JoRRP):

Old case definition: Please report any infant or child under the age of 15 years diagnosed with Juvenile onset Recurrent Respiratory Papillomatosis (JoRRP) confirmed by endoscopy of the larynx AND by histology."

New case definition: Please report any infant or child under the age of 15 years diagnosed with Juvenile onset Recurrent Respiratory Papillomatosis (JoRRP) confirmed by endoscopy of the larynx *with or without confirmation by histology.*

Please continue to report cases of JoRRP when they are seen.

INFLUENZA Study 2014

APSU will once again conduct national surveillance for Severe Complications of Influenza from July to September 2014.

Please report all children with laboratory confirmed influenza admitted to hospital with severe complications such as pneumonia requiring oxygen therapy, encephalitis and other neurological complications, rhabdomyolysis. The protocol and questionnaire are available on the APSU website www.apsu.org.au

APSU surveillance over the last 7 years has identified some of the more serious and unusual complications of influenza in children. A number of child deaths have also been reported to the APSU, including in previously healthy children.

Children with chronic conditions are especially vulnerable to influenza complications. Such children are recommended and funded under the National Immunisation Program for seasonal influenza Vaccination.

Look out for flu and report all serious cases to APSU!



Rare Disease Day Events

To coincide with International Rare Disease Day, APSU once again held an educational workshop for health professionals and families over two days 14-15 March 2014. The workshop was opened by A/Professor Susan Moloney, the President of the Division of Paediatrics and Child Health, Royal Australasian College of Physicians. The workshop was held at the Kids Research Institute at Westmead with approximately 60 health professionals attending. There were outstanding presentations and panel discussions involving experts in rare diseases and families living with rare disease.

On day 1 health professionals discussed recent developments in rare diseases research, policy, diagnosis, new advances in genetics, and the needs for improved health services for people living with rare chronic and complex conditions. A special highlight was a presentation from Siobhan Darby who told us about her journey as a young woman living with a rare condition and her experiences of transition from paediatric health services to adult health services.



Siobhan Darby shares her story

A panel discussion about transition services for young people living with chronic conditions proved lively and robust. It highlighted new initiatives including the recently established TRAPEZE transition service in NSW.

Day 2 of the workshop brought together families and support organisations for discussions centred on out-of-pocket medical expenses and equitable access to health and community support services. Ms Dianne Petrie provided a perspective on the role of peer support organisations and Ms Lyndal Douglas spoke about the important role of genetic counsellors in supporting families.



Speakers participating in Day 1 (L - R):
Dr Neil Hime, Prof John Christodoulou, Dr Marie Deverell, Dr Sandra Johnson, Ms Siobhan Darby, Ms Kate Dunlop, A/Prof Susan Moloney, A/Prof Yvonne Zurynski, Dr Felicity Collins, A/Prof Nigel Clarke, Prof Elizabeth Elliott

Interesting Findings and snippets

Congenital Rubella Syndrome

There were 3 cases of congenital rubella syndrome reported in recent years (2012 and 2013) after 5 years of no reports. All 3 children were born to immigrant mothers from countries where there are no routine rubella immunisation programmes.

Vitamin K deficiency bleeding (VKDB)

This is a rare condition in infants – APSU usually received none or one report per year. Since January 2013 there have been 6 reports – all infants born in 2013 and in 5 of the 6 vitamin K prophylaxis at birth was not given because parents refused consent.

Why continue surveillance for Acute Flaccid Paralysis?

Although Australia has been certified by the WHO as “polio-free” for many years poliovirus still circulates in some regions of the world. With high levels of travel and increased immigration from endemic countries, polio continues to be a threat. There has been a recent increase in the international spread of polio which prompted an alert issued by the WHO <http://www.who.int/mediacentre/news/statements/2014/polio-20140505/en/>

Travellers entering Australia after travel from the following countries are especially at risk: Pakistan, Cameroon, Syria, Afghanistan, Equatorial Guinea, Ethiopia, Iraq, Israel, Somalia and Nigeria
APSU was able to directly inform 1400 paediatricians of this alert.
Please continue to report cases of Acute Flaccid Paralysis to the APSU.

Congratulations

Dr Angela McGillivray and the APSU Severe Neonatal Hyperbilirubinaemia Study Group. Angela won the New Investigator Award at PSANZ Conference for her presentation entitled: PROSPECTIVE SURVEILLANCE STUDY OF SEVERE NEONATAL HYPERBILIRUBINAEMIA IN AUSTRALIA.

Recent publications

Anderson A, Wong K, Jacoby P, Downs J, Leonard H. Twenty years of surveillance in Rett syndrome: what does this tell us? *Orphanet Journal of Rare Diseases* 2014. 06/2014; 9(1):87. DOI:10.1186/1750-1172-9-87

Basha J, Iwasenko JM, Robertson P, Craig ME, Rawlinson WD. Congenital cytomegalovirus infection is associated with high maternal socio-economic status and corresponding low maternal cytomegalovirus seropositivity. *Journal of Paediatrics and Child Health*, 2014, 50(5):368-372

Hime NJ, Fitzgerald D, Robinson P, Selvadurai H, Van Asperen P, Jaffé A, Zurynski Y. Childhood interstitial lung disease due to surfactant protein C deficiency: Frequent use and costs of hospital services for a single case in Australia. *Orphanet Journal of Rare Diseases* 2014. 9(1), 19 Article number 36

Zurynski, Y., McIntyre, P., Booy, R., Elliott, E.J. Paediatric active enhanced disease surveillance: A new surveillance system for Australia *Journal of Paediatrics and Child Health*, 2013. 49(7): 588-594.

APSU Team Changes

Dr Neil Hime, Dr Yemisi Ijamakinwa and Dr Victor Wu have departed the APSU after completing their contracts. Neil, Yemisi and Victor contributed an enormous amount of work towards the Childhood Chronic Diseases Project funded by the Australian Department of Health. **Ms Jade Mangan**, Administration Officer has moved to take on a position with the Education Centre at the Children's Hospital at Westmead, and **Ms Kirrilee Drew**, APSU Office Manager has moved to take on the position of Executive Officer with The Kids Research Institute, Westmead.

We would like to thank Neil, Yemisi, Victor, Jade and Kirrilee for their hard work and dedication to the APSU.

A big thank you to volunteer student Charmy Fernando who assisted the APSU in the latter half of 2013.

APSU Staff Christmas Party 2013:

(L - R, Back Row)

Diana Thomas, Neil Hime, Ingrid Charters,
Yemisi Ijamakinwa, Marie Deverell, Jade Mangan.

(L - R, Front Row)

Kirrilee Drew, Tracey Tsang, Yvonne Zurynski,
Jennifer Johnson, Jocelynn McRae.

(Absent: Elizabeth Elliott, Amy Phu, Premala Sureshkumar,
Victor Wu)



Still returning the yellow card by post? We aim to have all contributors responding by email by the end of this year. If you are one of the remaining ~100 clinicians still receiving the yellow report card we would like to make responding easier for you. Please e-mail apsu@chw.edu.au or call 02 9845 3005 and let us know your work email and you can start responding to an e-mail version of the yellow card.

Not currently an APSU Contributor?

If you would like to become a member of the surveillance program please contact our office on 02 9845 3005 or apsu@chw.edu.au

Changed your contact details? Please let us know by contacting the APSU office on 02 9845 3005 or apsu@chw.edu.au to enable continued participation in the APSU.

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