

rare

Kids



Australian Paediatric Surveillance Unit **NEWSLETTER** MARCH 2015

New Studies - recently added to report card

Fetal Alcohol Spectrum Disorders (FASD)

FASD are lifelong but potentially preventable conditions. Consequences of in utero exposure to alcohol include brain damage, growth failure, birth defects and problems with development, learning, behaviour and mental health. This study, led by Professor Elizabeth Elliott, will allow us to estimate the incidence of fetal alcohol syndrome (FAS), partial fetal alcohol syndrome (PFAS), and neurodevelopmental disorders-alcohol exposed (ND-AE) in children <15 years. In addition it will allow us to describe current diagnostic practice, presenting features, use of, and gaps in health services for children with FASD. Please report all cases diagnosed from 1 December 2014 according to the protocol and questionnaire distributed to you and available on the APSU website www.apsu.org.au.

Childhood Interstitial Lung Disease (chILD)

The cause of chILD is often unknown but the disease may be a consequence of genetic mutation, environmental exposure or sequelae of systemic disease. The rarity of chILD disorders means that current clinical guidelines for diagnosis and management are based primarily on clinical experience and informed judgement, rather than systematic study.

This study, led by Professor Adam Jaffe, it will allow us to describe the frequency of chILD, demographics, presentation and diagnoses or short-term outcomes. Please report all cases diagnosed from September 2014 according to the protocol and questionnaire distributed to you and available on the APSU website www.apsu.org.au.

New Studies - to be added in the near future

- ◆ Chronic Fatigue Syndrome
- ◆ Early Onset Eating Disorders
- ◆ 22q11.2 Deletion Syndrome
- ◆ Obesity and Obstructive Sleep Apnoea

Do you have an idea for an APSU Study ?

We have had a number of expressions of interests regarding future APSU studies including:-

- ◆ Hypophosphatemia
- ◆ Non-Bacterial Osteomyelitis

If you have a study that you feel is suitable for the APSU, then we want to hear from you. Please contact our office or visit our [website](http://www.apsu.org.au) to obtain the Expression of Interest Form.

Recent publications

Desai S, Smith T, Thorley BR, Grenier D, Dickson N, Altpeter E, Sabbe M, Elliott E, Zurynski, Y. Performance of acute flaccid paralysis surveillance compared with World Health Organization standards. *Journal of Paediatrics and Child Health* 2014, 57 (2):209-214.

Hime NJ, Zurynski Y, Fitzgerald D, Selvadurai H, Phu A, Deverell M, Elliott E, Jaffe A. Childhood Interstitial Lung Disease: A Systematic Review. *Pediatric Pulmonology*. 2014. (In press)

Khandaker G, Zurynski Y, Ridley G, Buttery J, Marshall H, Richmond PC, Royle, J, Gold, M, Walls, T, Whitehead, B, McIntyre, P, Wood, N, Booy, R, Elliott, EJ. Clinical epidemiology and predictors of outcome in children hospitalised with influenza A(H1N1) pdm09 in 2009: a prospective national study. *Influenza Other Respir Viruses*. 2014. 8(6):636-45.

Khandaker G, Zurynski Y, Jones C. Surveillance for congenital rubella in Australia since 1993: Cases reported between 2004 and 2013. *Vaccine*. (0). 2014, 32(50):6746-6751.

Ko D.W.H., Zurynski Y, Gilbert GL, and members of the APSU GBS study group. Group B streptococcal disease and genotypes in Australian infants. *Journal of Paediatrics and Child Health* 2015. (in press)

Mackie FE, Kainer G, Adib N, Boros C, Elliott EJ, Fahy R, et al. The national incidence and clinical picture of SLE in children in Australia - a report from the Australian Paediatric Surveillance Unit. *Lupus* 2015, 24(1):66-73.

Macartney KK, Gidding HF, Trinh L, Wang H, McRae J, Crawford N, et al. Febrile seizures following measles and varicella vaccines in young children in Australia. *Vaccine*. 2015;33(11):1412-7.

Mehr SS, Campbell D, Joshi P, Smart J, Peake JE, Smith PK, et al. Fpies Epidemiology in Australia: Results from a 2-Year Prospective Population Study. *Journal of Allergy and Clinical Immunology*.135 (2):AB168.

The APSU Research Report is coming!

We are currently preparing the next APSU research report which will report on the period 2011–2014, and will be available later this year. To view previous APSU research reports please visit the [website](http://www.apsu.org.au).



RARE DISEASE DAY

The Steve Waugh Foundation Rare Star Event at Taronga Zoo, 25 February



APSU's Deputy Director, Yvonne Zurynski was an invited speaker at this event and discussed the frequent use of specialist health services among children who have a rare disease. The Rare Star event celebrated the support provided by the Steve Waugh Foundation to many families living with children who have a rare disease. It was a fun day for children and families with special prizes and surprises organised by the Steve Waugh Foundation and their major partner Canon Australia. The APSU continues to support the Steve Waugh Foundation through the involvement of Elizabeth Elliott and Yvonne Zurynski in the Medical Health Advisory Committee, and our partnership in the APSU ARC Linkage Project Grant. For more information about the Steve Waugh Foundation please go to <https://www.stevewaughfoundation.com.au/>.

APSU ARC Linkage Project Grant Update

"Psychosocial and Economic Impacts of Rare Diseases on Australian Children, Families and Health Professionals." (Yvonne Zurynski, Elizabeth Elliott, Helen Leonard, John Christodoulou).

Partner Institutions: Royal Australasian College of Physicians; Genetic Metabolic Disorders Service, The Children's Hospital at Westmead, Steve Waugh Foundation, SMILE Foundation (now part of Variety Australia), and Genetic Alliance Australia (formerly The Association of Genetic Support of Australasia).

The ARC linkage grant is now in its final year. The **survey of families** caring for a child with a rare disease was completed in 2014. We received 462 surveys (32% participation rate), with approximately 200 different rare conditions represented. Data analysis has been completed and we are currently preparing manuscripts on health service utilisation and the psychosocial and peer support needs of families.

The **audit of admissions and presentations** to the Genetic Metabolic Disorders Service at The Children's Hospital at Westmead is progressing steadily. Data on encounters and associated cost of service has been obtained from the Management Support Analysis Unit and the team are currently conducting the analyses with results to be published later this year.

The **paediatricians' survey** will highlight current awareness of rare diseases, need for information about referral pathways, diagnostic and clinical services accessed for rare disease patients, needs for educational materials and opportunities, and the preferred way to deliver these.

We have randomly selected ~600 APSU clinicians to participate in this important survey. You may have received an email from us asking you to complete a survey titled **"Paediatricians Caring for Children with Rare Diseases: Survey of current knowledge, practice and future educational needs."**

The survey takes approximately 15 minutes to complete and those who complete the survey will go into the draw to win a \$50 gift voucher. If you have been approached to participate in this study, please complete the online survey as soon as possible. If you have any questions regarding the survey, please contact us on 02 9845 3005 or email the APSU at apsu@chw.edu.au.

Other News

Information on rare diseases for parents and families

The APSU has started a collection of fact sheets on rare diseases which you may find useful for your patients, yourself or your colleagues. These are available from the Parents Portal on the APSU website (<http://apsu.org.au/home/parents/>). Recent additions include: Childhood Interstitial Lung Disease and Non-CF Bronchiectasis ([Children's Interstitial Lung Disease \(chILD\)](#)) and [Non-Cystic Fibrosis Bronchiectasis](#)). Don't forget that all APSU protocols past and present are on the APSU website under the Studies tab (<http://apsu.org.au/studies>). The protocols provide information about the rare conditions studied by the APSU.

APSU Scientific Review Panel

Sadly we recently received notification of Neil Wigg's resignation from the APSU Scientific Review Panel—***"My association with the APSU over the past decade has been very rewarding and I enjoyed working with the SRP and the Board. I quickly came to appreciate the research and educational value of the Unit, and I am sure its great contribution to children's health will continue."*** We would like to express our appreciation for Neil's involvement and his always excellent contribution to the activities of the APSU SRP and Board over the past decade, and wish him all the very best in his future endeavours.

Completed Studies/ Study Progress

Sudden Unexpected Early Neonatal Death or collapse (in previously healthy term infants in the first 7 days of life) (SUEND)

This study concluded in December 2014. There were 69 notifications of SUEND between January 2012 and December 2014. The investigators group led by Prof Heather Jeffrey 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 SUEND.

Female Genital Mutilation and Cutting (FGMC)

A big thank you to all APSU contributors who participated in this once-off study and survey which was the first study worldwide of paediatricians' knowledge, attitudes and practice related to FGMC in children. Approximately 10% of paediatricians who responded had seen at least one case of FGMC during their clinical career, over 50% knew of the WHO guidelines for FGMC and ~30% were aware of the RACP Guidelines on FGMC. Paediatricians called for education about treatment and prevention of FGMC. The study was conducted separately from the usual APSU report card from March to June 2014. Our thanks go to the steering committee who guided the study: Nesrin Varol, Karen Zwi, Susan Moloney, Shanti Raman, Jacqueline Small, Ajay Rane, Juliana Nkrumah, Elizabeth Elliott, Yvonne Zurynski, Marie Deverell, and to Premala Sureshkumar who coordinated the study.

Severe Complications of Influenza

This study was once again conducted from July to September 2014 and data has now been analysed. Thank you to all clinicians for contributing cases to this study. Key findings were as follows:-

There were 81 cases reported of children admitted to hospital with serious complications of influenza (76 Flu A; 4 Flu B; 1 Flu A & B) including pneumonia, encephalitis, seizures and rhabdomyolysis. 29 children required an ICU admission and one child died. Of the 81 children, 67.9% were previously healthy, while 32% had chronic pre-disposing conditions including neuromuscular disorders, cerebral palsy, asthma, chronic lung disease, Rett syndrome, CHARGE and SCN1B mutation. Only 5 children were vaccinated for influenza within the last 12 months. Children with chronic predisposing conditions are recommended and funded for annual influenza vaccination under the National Immunisation Program (<http://www.immunise.health.gov.au/internet/immunise/publishing.nsf/Content/immunise-influenza#flu>).

Congenital and Neonatal Varicella

Surveillance for congenital and neonatal varicella has been on-going since 2006. There has been a significant decline in the number of cases reported over the years and since the introduction of varicella vaccination into the National Immunisation Program as shown by our paper published in 2011 (Khandaker G, Marshall H, Peardon E, Zurynski Y, Burgner D, Buttery J, et al. Congenital and neonatal varicella: impact of the national varicella vaccination programme in Australia. *Arch Dis Child*. 2011;96(5):453-6). The incidence of congenital and neonatal varicella has continued to decrease since then and the investigators plan to publish an update on surveillance data covering the period 2006 to 2014. **Please report any outstanding cases of congenital or neonatal varicella as soon as possible and provide any outstanding case report forms for already reported cases.**

RedCap Online Reporting Clinicians are now able to complete most case report forms online through our data capture management system (RedCap). Simply click on the link provided in the monthly report card email to complete the form. Links to the current APSU protocols and case report forms can be found in the monthly email report card and are also available for download on the APSU website. If you have any difficulties accessing the links or would like more information about completing the forms online, please contact us.

Still returning the yellow card by post? If you are one of the remaining 113 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 programme 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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