rare kids

Australian Paediatric Surveillance Unit NEWSLETTER August 2013

Welcome to the July edition of the Rare Kids Newsletter. We hope you enjoy this edition and as always, welcome any comments or feedback you may have.

Have you registered yet?

There is less than 4 weeks until the 27th Congress of the International Pediatric Association. Our pre-congress workshop on Saturday 24th August will showcase the achievements of the International Network of Paediatric Surveillance Units (INoPSU) over the past 15 years, including the impacts of results on clinical practice and policy. Esteemed international speakers including Odille Kremp, France; Elizabeth Elliott, Australia; Danielle Grenier, Canada; Christopher Verity, United Kingdom; Sloane Madden, Australia; Yvonne Zurynski, Australia; and Nigel Dickson, New Zealand will be presenting. Please visit http://www2.kenes.com/ipa/scientific/Pages/Workshops.aspx. See the detailed program on the last page of the newsletter.

A session dedicated to Rare Diseases will be held on Sunday 25th August from 11.00-12.30. Guest speakers Dr Odile Kremp, France; Prof Chris Verity, UK; and Dr Danielle Grenier, Canada will be featured in the session. We will be launching the APSU 20 Year Report at the Sunday session.

New Studies

Flu season is upon us

The APSU will once again conduct our **seasonal flu surveillance** for the Department of Health and Ageing. Surveillance commenced on 1 July 2013 for the severe complications of laboratory confirmed influenza in children aged < 15 years and admitted to hospital. Surveillance will conclude on 30 September. Please report any child with severe complications of laboratory proven influenza and admitted to hospital as soon as they are seen. Please see our website for more details—www.apsu.org.au

EoE—Eosinophilic Eosophagitis

Eosinophilic Eosophagitis (EoE) is a non-IgE mediated chronic allergic disorder which appears to be triggered, in many cases, by ingestion of certain food protein(s). Please report any child < 16 years of age newly diagnosed with EoE, whom you have seen within the last month and that you have not previously reported to the APSU. **Case definition criteria**:

Demonstrated increased numbers of eosinophils (>15 per high power field) in ***at least one*** oesophageal biopsy (lower, mid or upper).

AND Excluding Gastro-oesophageal Reflux Disease which has responded to a Proton Pump Inhibitor. For full condition description and definition please visit our website www.apsu.org.au/current studies

APSU Studies

Please find below a complete list of current studies under surveillance with the APSU

- Severe Complications of Influenza
- Eosinophilic Eosophagitis (EoE)
- Sudden Unexpected Early Neonatal Death or Collapse
- Food Protein Induced Enterocolitis Syndrome
- Juvenile onset Recurrent Respiratory Papillomatosis
- Congenital varicella
- Neonatal varicella
- Severe complications of varicella#
- Rett syndrome
- Congenital cytomegalovirus infection
- Newborn and infant herpes simplex virus infection
- Acute flaccid paralysis
- Paediatric HIV infection OR perinatal exposure to HIV
- Vitamin K deficiency bleeding (includes haemorrhagic disease of the newborn)
- Congenital rubella

New Management of Paediatric HIV infection study

The HIV infection and perinatal exposure to HIV study is now being managed by the APSU team and we would ask that all cases are reported directly to the APSU office and that all questionnaires are directed to our office.

The questionnaire for both the Mother and Child have been updated to reflect this management change and are available for download from our website http://www.apsu.org.au/studies/current/

Current studies update

Food Protein Induced Enterocolitis Syndrome (FPIES)

A new questionnaire has been developed for the FPIES study and is available for download from our website www.apsu.org.au. We kindly ask that all clinicians use this new form for reporting FPIES in future. The new questionnaire will allow our surveillance to better capture more detailed information regarding the child's diet at the time of reaction in order to better describe any cross-reactivity between food triggers causing FPIES.

The additional information sought will aid in determining whether children with cow's milk or egg FPIES have managed to tolerate baked cow's milk or egg products.

Congenital Rubella Syndrome—please report all cases seen

Rubella infection *in-utero* can lead to Congenital Rubella Syndrome (CRS) which is characterised by significant birth defects and other problems including: cataracts, microophthalmia, glaucoma, pigmentary retinopathy, sensorineural deafness, heart defects, and neurological problems including microcephaly, psychomotor retardation, behavioural disorders. Long term effects include subacute sclerosing panencephalitis (SSPE) and diabetes mellitus.

In recent years there have been no reports of new cases of CRS to the APSU, however we remain vigilant. The recent measles outbreaks suggesting reduced MMR coverage, and increased immigration from countries with poorly established rubella vaccination programs may result in increased CRS cases.

It is important to notify all cases of Congenital Rubella so that they can be described in detail, and missed opportunities for prevention identified and addressed. Please report any cases seen, including any cases already reported to Public Health Authorities, to the APSU as soon as possible.

Transition to adult services—trials and tribulations

A transition forum held on the 23rd of February at the University of Sydney, identified gaps and needs for young people living with complex, chronic rare diseases and medical conditions.

Young people and their parents called for enhanced transition services with adequate preparation for transition, better linking between paediatric and adult health services and a "One Stop Shop" where they could access a case manager or "transition coach" who could help them navigate the transition process, including dealing with agencies such as Medicare and private health insurance providers. The report describes these issues from the young person's point of view. An editorial on this subject was published by MJA; https://www.mja.com.au/journal/2013/198/11/challenges-transition-adult-health-services-patients-rare-diseases

The future for Rare Diseases

The Sydney Children's Hospital Network (SCHN) has recently published a draft strategic plan for 2013-2017 in which Rare Disease Research has been identified as a priority area. This will encourage innovative approaches to drive and achieve excellence in rare diseases and genomics. To read the plan http://intranet.schn.health. nsw.gov.au/research/draft-research-strategic-plan-2013-2017

Living with Rare Diseases

Under the ARC Linkage Grant we have developed a detailed questionnaire to capture the impacts on children and their families who are affected by a rare disease. The 16-page questionnaire focuses on diagnosis, health related functions, treatment, health service use, family impact, support and information needs and financial support.

Newest recruits to the APSU

The APSU would like to welcome Dr Neil Hime (Research Associate) and Dr Oluyemisi (Yemisi) Ijamakinwa (Research Assistant) to the APSU Research Team.

Neil has a Masters of Public Health and is a medical and public health researcher with extensive experience directing projects across a number of medical fields, and has a strong interest in the socio-economic impacts that contribute to the growing burden of disease in Australia.

Yemisi has an MBBS as well as a Masters degree in Public Health and Health Management. Her extensive background in clinical medicine and public health, coupled with her strong interest in research, makes her a valuable resource to APSU projects.

Both Neil and Yemisi bring with them an excellent specialised skill set which will be very beneficial to the APSU and the work we conduct.

Please join us in welcoming Neil and Yemisi to the APSU Research Team.

Recent Publications

Esterman EE, Lahra MM, Zurynski YA, Booy R, Elliott EJ. Influenza infection in infants aged <6 months during the H1N1-09 pandemic: a hospital-based cohort study. Journal of Paediatrics and Child Health 2012: June 18: DOI: 10.1111/jpc.12266 (online first)

Zurynski Y, McIntyre P, Booy R, Elliott EJ, for the PAEDS Investigators Group. Paediatric Active Enhanced Disease Surveillance (PAEDS): a new surveillance system for Australia. Journal of Paediatrics and Child Health 2012; 49 (7):588-594.

Anderson M, Elliott EJ, Zurynski YA. Australian families living with rare disease: experiences of diagnosis, health services use and needs for psychosocial support. Orphanet Journal of Rare Diseases. 2013;8 (1):22. Epub 2013/02/13.

Zurynski Y, Elliott EJ. Challenges of transition to adult health services for patients with rare diseases: Med J Aust 2013; 198 (11): 575-576

The questionnaire will be a valuable tool to gain insight into what it really means to live with a rare disease. We would like to thank our partner organisations for their contribution and ongoing support to this research project: The Steve Waugh Foundation, SMILE Foundation, AGSA, The University of Sydney and The Sydney Children's Hospital Network.

Director of Orphanet visits Australia

Segolene Ayme, the Director of Orphanet, visited Perth in May. Dr Ayme's visit was supported by the Office of Population Health Genomics at WA Health. She presented a seminar at Murdoch University on rare diseases. the need for incidence and prevalence data, rare disease databases and registries, and the need for practical information for clinicians.

The seminar was attended by a mix of academics, clinicians, policy makers and representatives from the rare diseases community. Yvonne Zurynski and Elizabeth Elliott met with Dr Ayme to discuss potential collaboration between APSU, INoPSU and Orphanet – we look forward to further developing these collaborations.

Orphanet is a web portal providing summary information about rare diseases which is easily available to clinicians and patients and includes a "search by symptoms" facility as well as ability to search by disease. They also provide access to clinical guidelines, lists of rare disease clinical trials, clinics and parent supports.

An Orphanet Australia Committee has been established and both Yvonne and Elizabeth will serve on this committee.

You will find a wealth of information at the Orphanet site:
http://www.orpha.net/consor/cgi-bin/index.php

Rare diseases debate begins in Parliament

A debate about rare diseases has begun in Canberra, with key organisations including the APSU mentioned in the debate. Rare disease advocacy groups and foundations including Rare Voices Australia have been working very hard to advocate on behalf of the rare disease community and to engage with parliamentarians to bring attention to the common problems of rare diseases.

Link to the Hansard:
http://www.openaustralia.org/debates/?id=2013-06-24.186.1&s=australian+paediatric+Surveillance+Unit#g190.1

Changed your details?

To ensure we do not lose touch, please contact our office and advise us of any changes to your contact details.

Useful links to rare diseases advocacy groups and Foundations:

Rare Voices Australia
http://www.rarevoices.org.au/

SMILE Foundation
http://www.smilefoundation.com.au/

Steve Waugh Foundation
http://www.stevewaughfoundation.com.au/

Association of Genetic Supports Australasia
http://www.agsa-geneticsupport.org.au/

Keep your eyes peeled

We are delighted to announce that our very own Dr Sandra Johnson has recently published her book for parents entitled "Your Child's Development—Nurturing healthy self esteem and relationships". The book provides a practical approach to child rearing and will prove to be a helpful resource for anyone working with children and adolescents. For more details about the book and to purchase the book please visit http://www.amazon.com/dp/B00CFA0HBK.

Not currently an APSU Contributor?

The APSU currently has more than 1390 paediatricians participating in our surveillance program.

If you would like to join the surveillance program and become more involved with the APSU simply contact our office on 02 9845 3005 or apsu@chw.edu.au and we will do the rest.

The APSU Team

International Network of Paediatric Surveillance Units: The Power of International Collaboration to Study Rare Diseases

Saturday 24th August 2013 0830-1430

This unique workshop brings together research into rare childhood conditions from around the Globe. The International Network of Paediatric Surveillance Units comprises 12 units from 12 countries around the world, providing a powerful means of collecting international data on many rare conditions simultaneously. Learn about the innovations that have enabled INoPSU to influence clinical practice and public health policy. Make new international connections and find out how to establish a paediatric surveillance unit in your country.

Time	Topic	Speaker(s)
0830	*Coffee, pastries and welcome*	
Session Chair: Yvonne Zurynski		
0900-0930	Public health impacts of INoPSU data – international perspectives	Danielle Grenier Canada
0930-1000	Haemolytic Uraemic Syndrome: international comparisons in epidemiology	Elizabeth Elliott Australia
1000-1030	Need for data and registries for rare diseases – Latest developments in Europe	Odille Kremp, France ORPHANET
1030-1100	*Coffee break*	
Session Chair: Alan Emond		
1100-1130	Early Onset Eating Disorders – international comparisons in children aged < 13 years	Sloane Madden Australia
1130-1200	Preventing serious injuries in children – using evidence to change policy	Yvonne Zurynski Australia
1200-1230	Progressive intellectual and neurological deterioration in children: a complex mixture of rare conditions	Christopher Verity England
1230-1330	***Lunch*** *and Launch of the 15 year INoPSU Report and new website – Yvonne Zurynski and Danielle Grenier*	
Session Chair: Elizabeth Elliott		
1330-1400	Hyperbilirubinaemia- international perspectives	Matthias Roth Switzerland
1400-1430	NZPSU: impacts on clinical practice and policy in New Zealand	Nigel Dickson New Zealand
1430	*End of session*	
1430-1745	INoPSU Business Meeting **INoPSU Members ONLY from 1430**	
1800	Congress Opening Ceremony	
2000	INoPSU Dinner	

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