



## Australian Paediatric Surveillance Unit

# Rare Disease Workshop

**“Recent developments in rare diseases research, health services and policy”**

**You MUST register  
to attend – limited  
spaces available!!**

**Friday 14<sup>th</sup> March 2014; 9am to 4pm  
Kids Research Institute  
Children’s Hospital at Westmead  
Cnr Hawkesbury Rd & Hainsworth St  
Westmead NSW 2145**

This workshop is designed for health professionals, policy makers and researchers. Under the theme **“Recent developments in rare diseases research, health services and policy”**, the programme will include sessions on the diagnosis of rare diseases including the latest on genetic testing; the psychosocial impacts of rare diseases on families; health services utility and needs; and the role of rare disease registries to support research and clinical care. Please see the programme on the next page.

**Cost: \$50.00** includes lunch, morning and afternoon tea

\*Trainee health professionals and students in the Sydney Children’s Hospitals Network – NO CHARGE but you must register as numbers are limited.

**REGISTRATION FORM** - To register please complete the form below or call the APSU on (02) 9845 3005

<b>Name:</b>		
<b>Your profession or job title:</b>		
<b>Address:</b>		
<b>Phone:</b>	<b>Mobile:</b>	<b>Email:</b>
<b>Your current position and place of work:</b>		
<b>Dietary requirements:</b>		
<b>*I am a trainee health professional or student in the Sydney Children’s Hospital Network</b> <input type="checkbox"/> <b>(no charge applies)</b>		

**Registration Fee \$50.00 (including GST)**

☐ I enclose a cheque for \$50.00 (Cheque to be made payable to the Australian Paediatric Surveillance Unit - SCHN)

☐ Debit my credit card for \$50.00    Credit Card Type    Visa ☐    MasterCard ☐

Credit Card Name \_\_\_\_\_

Credit Card Number \_\_\_\_\_

Expiry Date \_\_\_\_\_    Signature \_\_\_\_\_



Please return completed registration forms to the **APSU** either via fax on (02) 9845 3082 or by email at [apsu@chw.edu.au](mailto:apsu@chw.edu.au)



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### Programme

Time	Title	Speaker
0900	Welcome	Prof Susan Moloney
0910-1030	<b>Session 1: Responding to the impacts of rare diseases – Chair Prof Susan Moloney</b>	
0910-0930	Responding to Rare Diseases – Recent Developments in Australia and Internationally	Prof Elizabeth Elliott
0930-0950	Psychosocial burden, health services use and needs among families living with rare disease	A/Prof Yvonne Zurynski
0950-1010	The cost of rare diseases – Health services use and out of pocket expenses	Dr Marie Deverell
1010-1030	Legal and social rights of families living with rare disease and disability	Dr Sandra Johnson
1030-1100	Morning Tea	
1100-1230	<b>Session 2: Diagnosis of rare diseases – Chair TBA</b>	
1100-1130	Update on genetic testing – who should we test and how?	Dr Felicity Collins
1130-1140	Finding a diagnosis for a very rare disease	Movie screening
1140-1210	Latest developments in gene discovery and clinical research to support diagnosis, guidelines and care	Prof Nigel Clarke
1200-1230	Where to go when you don't know: Educational resources on genetic disorders and rare diseases	Ms Kate Dunlop
1230-1330	Lunch	
1330- 1430	<b>Session 3 – health services for rare diseases – Chair A/Prof Yvonne Zurynski</b>	
1330-1335	Children's Interstitial lung Disease – Sarah's Story	Movie screening
1335-1340	Use of hospital services for Children's Interstitial lung disease – a case study	Dr Neil Hime
1340-1400	<i>Panel discussion</i> – Do we have adequate health services for children with rare chronic lung disease?	Panel: Prof Adam Jaffe, Prof Hiran Selvadurai, Dr Neil Hime
1400-1410	What's it like being a teenager with Klippel-Trenaunay Syndrome and moving to adult health services	Ms Siobhan Darby
1410-1430	<i>Panel Discussion</i> – transition services for young people living with rare disease	Ms Lynne Brodie, Ms Madeleine Bridget, Ms Sarah Ryan, Dr Marie Deverell
1430-1500	Afternoon Tea	
1500-1600	<b>Final session: Registries and surveillance and summing up - Chair Prof Elizabeth Elliott</b>	
1500-1530	Rare disease registries and rare disease surveillance to support research and policy	A/Prof Yvonne Zurynski and Prof Adam Jaffe (15 minutes each)
1530-1600	Panel Discussion – Raising the profile of rare diseases (panel to include researchers, clinicians, advocacy groups and family support organisations)	Prof Elizabeth Elliott to lead discussion
1600	Close	



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