

### Background

Rare diseases affect a small number of people relative to the general population and specific issues are raised in relation to their rarity. In Europe for a disease to be considered rare it must affect 1 person per 2,000. A disease can be rare in one region, but common in another. Furthermore, there are many common diseases whose variants are rare.

The field of rare disease research suffers from a deficit of medical and scientific knowledge. For a long time awareness of rare diseases was scarce and until very recently there was no real research or public health policy concerning issues related to the field.

Researchers and other stakeholders are increasingly working through networks to share the results of their research, advancing scientific knowledge more efficiently. Networks of registries and surveillance systems are effective instruments to improve and disseminate knowledge on rare and uncommon diseases and conditions. Surveillance units have been widely used in the field of paediatrics and child health, as well as in other settings.

The International Network of Paediatric Surveillance Units (INoPSU), was established in 1998 and involves 12,000 paediatricians covering a child population of 50 million. It currently has 12 member units (Australia, Belgium, Canada, Germany, Greece, Netherlands, New Zealand, Portugal, Ireland, Switzerland, UK, and Wales).



### Aims

This presentation describes the impact of data collected by paediatric surveillance units (PSUs), both individually and collectively as members of INoPSU, on public health outcomes, clinical care and research. Highlighting the strength of this active surveillance methodology.

### Methods

All PSUs use a surveillance method based on systematic, active, individual reporting to facilitate case collection of rare childhood conditions - including infectious and non-infectious disease and mental health issues.

PSUs provide a structured but flexible system for researchers to communicate with practising paediatricians and other child health specialists to collect data.

Each month a report card listing the conditions under surveillance is sent nationally to paediatricians; both negative and positive reports are collected. Positive reports are followed by a data collection form. Post, e-mail or the Internet is used for case reporting and data collection. Confidentiality of data is required. The use of additional sources of case ascertainment is strongly encouraged.

INoPSU acts as a virtual network through its website and e-newsletters. It helps facilitate the sharing of information and multi-national collaboration through the use of shared protocols and data.

### Results

INoPSU has facilitated surveillance of over 300 rare paediatric conditions, including over 70 rare infections.

The surveillance projects have enabled researchers to:

- monitor public health interventions (effectiveness of immunisation programmes)
- characterise the epidemiology of emerging/re-emerging diseases and conditions (variant CJD, H1N1, ZIKA)
- inform the development of new screening policy (invasive group B Streptococcal disease, HIV, MCADD)
- identify geographical variations (haemolytic uraemic syndrome)
- describe the epidemiology of child mental health disorders and other childhood disabilities (eating disorders - anorexia nervosa)
- guide clinical practice policy (Vitamin K deficiency bleeding)

These projects had important impacts, surveillance of:

- acute flaccid paralysis (AFP) in Australia was central to the process of WHO certification of the Western Pacific region as polio-free
- congenital rubella, subacute sclerosing panencephalitis, meningoencephalitis, AFP and other conditions have been crucial to monitoring the effectiveness and safety of immunisation programmes
- childhood HIV surveillance led to changes in antenatal screening policy in Australia and the UK
- neonatal herpes simplex virus suggests the need for an HSV-1/HSV-2 effective vaccine
- injuries related to the use of baby-walkers and to the use of seat-belt provided evidence for legislative action in Canada and Australia.

Many papers have been published in peer-reviewed journals and informed national and international guidelines.

### Conclusions

Research networks are effective instruments to improve knowledge on rare and uncommon diseases and conditions. International collaboration between PSUs sharing interests, definitions and methods further improves the effectiveness of research efforts.

### What does this add to European paediatric research?

National PSUs and their International Networks are widely available and have proven to be useful and effective partners for research on rare and uncommon diseases and conditions.

### Selected joint papers

Grenier D, Lynn R, Zurynski Y on behalf of all national paediatric surveillance unit investigators. Public health impacts of the International Network of Paediatric Surveillance Units. *Paediatr Child Health*. 2009; 4(8):499-500.

Desai S, Smith T, Thorley BR, Grenier D, Dickson N, Altpeter E, SPSU Committee, Sabbe M, Elliott E, Zurynski Y. (2015). Performance of acute flaccid paralysis surveillance compared with World Health Organization standards. *Journal of Paediatrics and Child Health*. 2015, 51(2): 209-214.

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