Rare respiratory diseases in children
- impact on health professionals and health services

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Introduction

The French originally coined the term 'Orphan' disease, now defined as a disease with prevalence less than 1 in 2000 individuals. Rare diseases impose a significant burden on affected individuals, their carers, health providers, health resources and the community. It is estimated that there are 7000 rare diseases affecting approximately 50 million people in Europe and North America. Many of these are respiratory diseases.

Because of their rarity and the disparate location of patients throughout Australia, physicians may have never seen a patient with a particular Orphan disease. Thus often there is a delay in diagnosis and patients may undergo multiple unnecessary tests. When the diagnosis is ultimately made, the physician may not have any experience in managing patients with the disease. Furthermore, there may be little evidence based medicine to guide best practice. Patients may be treated with experimental drugs in an uncontrolled research setting. Furthermore, there may be little advice on the prognosis of the disease which is extremely frustrating for patients. Patient support groups may also be unavailable, leaving the patient and their family feeling helpless.

Rare respiratory diseases, like other rare diseases, may begin in childhood and be lifelong. The economic impact on the health care system is not insubstantial. What is needed is a clear coordinated collaborative approach. Patients with Orphan diseases need to be identified throughout Australia (For Respiratory Orphan diseases see: www.arnold.org.au) and Centres of Excellence established. These centres will help coordinate care via a multidisciplinary team approach (in partnership with paediatricians and adult physicians), develop local standards of care, enable research and promote education which should begin at University level and be part of the health professionals' curriculum. Furthermore, a coordinated approach will empower patients to establish support groups. These groups have a strong history in other countries for highlighting the importance of Orphan diseases to Governments and have resulted in the establishment of specific organisations to fund rare disease initiatives. This approach will improve health outcomes and reduce healthcare costs.

The French National Plan for rare diseases (2005-2008) represents an excellent framework for addressing the problems associated with rare lung diseases. Their objectives centre around strategic priorities as follows: to increase knowledge of the epidemiology of disease; to recognise the specificity of these diseases; to develop information for patients, health professionals and the general public; to train health professionals in identifying rare diseases; to organise screening and access diagnostic tests; improve access to treatment and inequality of patient care; support orphan drug development; respond to the specific needs of patients suffering from rare diseases; to promote research and develop national and European partnerships. Furthermore the French have made it a legal requirement that those patients with a rare disease get equitable treatment.

Australia is one of the few developed countries who do not have a National Plan. This urgently needs to be addressed.

Reference:

Jaffé A, Beville L, Zrynski Y, Elliott E. Call for a national plan for rare disease. *Journal of Paediatrics and Child Health*- 2009 Nov 23. [Epub ahead of print]