



Rare respiratory diseases in children – impacts on health professionals and health services

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Rebecca's Frustrations

- **Diagnosis**- lack of knowledge by physicians
- **Treatment**- experimental
- **Prognosis**- patients in limbo
- **Rules for equipment** e.g. home oxygen
- Lack of a **National Plan**

Intro



- We do little for ‘Orphan’ diseases
- Money
 - Drug companies
 - Research
 - No grants
 - Insufficient study power

The problem

- <1 in 2000 (European Parliament 1999)
- Approx 8000 rare diseases
- Approx 10% world has a rare disease

Zurynski MJA 2007

- Many of these are respiratory

Common features

- Low prevalence but high impact
- Many have childhood onset, lifelong, no cure
 - 20 year survival for at least one chromosomal abnormality 85.5% **Tennant et al Lancet 2010**
- Impact on patients/families
- Impact on medical health resources

Patients have the right to

- Equitable
 - prevention
 - diagnosis
 - treatment
- French Law

Impacts on Clinicians and Health Services

- Rett and Fetal Alcohol Syndrome
- Literature review
- Questions:
 - Impact on families and communities
 - impact on clinicians and health resources
 - How have governments responded

Impacts on Clinicians

- Lack of awareness of disease
 - Diagnostic delays
 - Wrong diagnosis
 - Multiple, unnecessary investigations
- Lack of evidence based information
 - Management guidance
 - Prognosis
- Lack of specialised diagnostic and referral services
- Lack of parent support groups

Impacts on Clinicians

- Unclear where to get information from
- Internet use
 - 46% NZ GPs used internet

Cullen RJ. *J Med Libr Assoc* 2002; 90:370-379.

- How many health professionals know about the Internet portals such as www.Orpha.net or Office of Rare Disorders

Impacts on Health Services

- Uncoordinated care
- Health expenditure
 - accessed frequently
 - specialised
 - disparate services
 - drugs
 - Fetal alcohol syndrome costs \$2.8 billion/year

Harwood H. National Institute on Alcohol Abuse and Alcoholism.

<http://pubs.niaaa.nih.gov/publications/economic-2000/#table3>

Impacts on Health Services

- Scarce resources
 - staff
 - equipment
 - clinic time
 - expertise
- Access to specialised clinics difficult
 - Rural Australia poses unique challenges
- Transition
 - Adult physicians experiencing new diseases

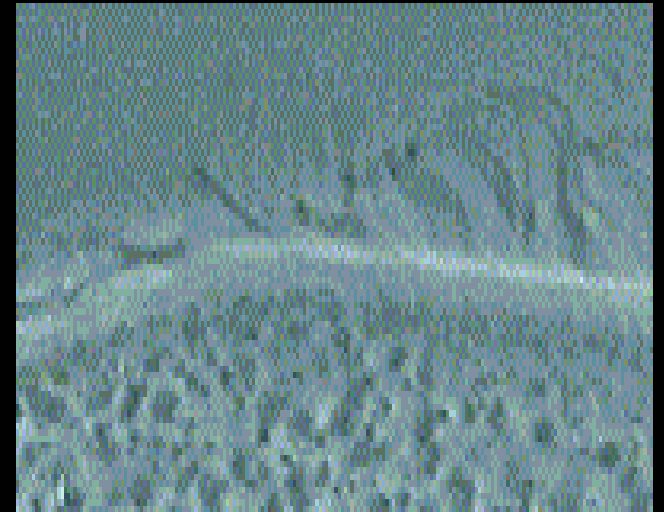
Poorer health outcomes

Patient outcomes

Health economic outcomes

Primary Ciliary Dyskinesia

- Ciliary abnormality
- Otitis media and deafness
- Bronchiectasis and respiratory failure
- Infertility



Primary Ciliary Dyskinesia

- 1 in 15 000
- 20 babies born a year
- Should be 1000 patients in Australia
- Diagnosis made by specialist techniques
 - nasal nitric oxide
 - microscopy

Ciliary Diagnostic Disorders in Australia

- 1 specialised centre
- No video EM
- Relies on 1 person
- Equipment bought by research grants or fund raising
- UK diagnostic centre exists

Ciliary Diagnostic Disorders in Australia

- Since 2000
- 500 referrals from all States
- 50 diagnosed
 - 1 have 1 patient in Bankstown
 - 1 in Wagga Wagga
- Where are the other 950?

Primary Ciliary Dyskinesia

The Solutions

- A National Centre of Excellence
- Similar to UK NSCAG model
- National referral centre
- Accurate diagnosis
 - State of the Art diagnostic facilities
 - No late diagnosis
- Establish local standards of care and benchmarks

Barbato et al ERJ 2009

Primary Ciliary Dyskinesia

The Solutions

- A National Centre of Excellence allows
 - Coordination of services
 - Equitable access
 - Paediatric and adult multidisciplinary team
 - NSW Kids
 - Transition
 - At little extra cost with improved health outcomes

The Solution

- Training
 - Medical students
 - Allied health
 - Nursing
 - Colleagues
- Sharing of expertise
 - Education to other hospitals
- International audit
- Research
- Patient/Family support group

National Registries



British Paediatric Orphan Lung Diseases (BPOLD)

THE DISEASES | MONTHLY RETURNS | NEWS | CONTACTS | LINKS | PATIENT & PARENT
FORUM | REGISTER

Welcome to BPOLD

BPOLD aims to establish a registry of nine rare lung diseases in children in the UK. The registry will provide epidemiological data on the prevalence and incidence of individual rare lung diseases and inform research projects which will increase our understanding of these diseases and ultimately improve treatment strategies for these children.

www.bpold.co.uk

Orphan Paediatric Lung Conditions

Click on the disease you require more information about. You can download the Acrobat

Disease	Date updated	Acrobat .PDF
Congenital Cystic Lung Lesions	11-11-2004	.PDF
Congenital Central Hypoventilation Syndrome	03-11-2004	.PDF
Interstitial Lung Disease	20-09-2004	.PDF
Obliterative Bronchiolitis	30-11-2004	.PDF
Pulmonary Alveolar Proteinosis	Soon	
Pulmonary Papillomatosis	19-11-2004	.PDF
Bronchiectasis of Unknown Cause	23-09-2004	.PDF
Pleural and Pulmonary Lymphangiectasia	06-02-2005	.PDF
Idiopathic Pulmonary Haemosiderosis	30-11-2004	.PDF



Estimated incidence patients under 16 in the UK, Aug 2006 to July 2007

Condition	Number reported Phase 1	Number reported Phase 2	Incidence per 1000,000
Congenital Cystic Lung Lesions	1	53 (5)	4.9
Congenital Central Hypoventilation Syndrome	0	10 (1)	0.9
Interstitial Lung Disease	2	15 (1)	1.4
Obliterative Bronchiolitis	7	14 (4)	1.3
Pulmonary Alveolar Proteinosis	0	3	0.3
Pulmonary Papillomatosis	0	3	0.3
Bronchiectasis of Unknown Cause	15	23 (3)	2.1
Pleural and Pulmonary Lymphangiectasia	1	6	0.6
Idiopathic Pulmonary Haemosiderosis	0	5	0.5

Laverty, Jaffe, Cunningham Ped Pulm 2008

Australasian Registry Network for Orphan Lung Disease

- To build a database of patients with rare lung diseases for future research
- Allow calculation of prevalence/incidence
- Conduit for research projects
- Reference portal for health professionals and patients
 - As an educational resource to improve clinical knowledge
 - Aid establish self-help groups

Methods

www.arnold.org.au

Australasian Registry Network for Orphan Lung Disease

[HOME](#) [MONTHLY RETURNS](#) [NEWS](#) [WHO WE ARE](#) [CONTACTS](#) [LINKS](#) [CLINICAL TRIALS](#) [PATIENT FORUM](#)

Orphan lung diseases

Congenital malformations

- Trachea
- Tracheo-oesophageal fistula
- Diaphragmatic hernia
- Cystic lung lesions
- Lobar emphysema
- Pleural/Pulmonary lymphangiectasia

Grown up (>16 years) chronic neonatal lung disease

Lymphangioleiomyomatosis

Alveolar haemorrhage syndrome

- Goodpasture syndrome
- Idiopathic pulmonary hemosiderosis
- Hereditary hemorrhagic telangiectasia (HHT) / Osler Weber Rendu (OWR) syndrome

Langerhans cell histiocytosis

Pulmonary vasculitides

- Wegeners granulomatosis
- Churg-Strauss vasculitis
- Other vasculitides incl. microscopic polyangiitis



Australasian Registry Network
for Orphan Lung Disease

Welcome to ARNOLD. The Australasian Registry Network for Rare Orphan Lung Disease.

Please use the menus on the left and right to access pages about the Adult and Paediatric Orphan Lung Diseases covered by this project.



ARNOLD - funded by the The Australian Lung Foundation

Orphan lung diseases

Bronchiolitis (in adults)

- Obliterative bronchiolitis (not transplant related)
- Diffuse panbronchiolitis
- Follicular bronchiolitis

Cryptogenic organising pneumonia

Hypereosinophilic lung diseases

- Chronic idiopathic eosinophilic pneumonia
- Acute idiopathic eosinophilic pneumonia
- Drug reactions with eosinophilia
- Loeffler syndrome

Extrinsic allergic alveolitis

Children's interstitial lung disease (ChILD)

Pulmonary amyloidosis

Pulmonary alveolar proteinosis

Pulmonary papillomatosis

Primary ciliary dyskinesia

Congenital central hypoventilation syndrome

Diseases

- 30 orphan lung diseases
 - disease information and web links being populated

ARNOLD-Australasian Registry Network for Orphan Lung Disease

References

Stocker J. Congenital and developmental diseases. In: Dail DH, Tomashefski JF, Cagle P, Hammar SP, Farver C, Colby TV, et al., editors. Dail and Hammar's Pulmonary Pathology. 3rd ed. New York: Springer-Verlag; 2008. p. 132-5.

Langston C. New concepts in the pathology of congenital lung malformations. *Semin Pediatr Surg.* 2003;12(1):17-37

Laberge JM, Puligandla P, Flageole H. Asymptomatic congenital lung malformations. *Semin Pediatr Surg.* 2005;14(1):16-33

Davenport M, Warne SA, Cacciaguerra S, Patel S, Greenough A

Congenital malformations - Cystic lung lesions

Dr Sandra Chuang Respiratory fellow Sydney Children's Hospital, Randwick, Australia


Definition: Congenital cystic lung lesions encompass a wide spectrum of rare lung malformations including congenital pulmonary airway malformation (CPAM) (previously termed congenital cystic adenomatoid malformations (CCAMs)), bronchopulmonary sequestration (intralobar and extralobar), bronchogenic cysts, and congenital lobar emphysema (CLE, also known as congenital lobar overinflation). These congenital malformations of the developing lungs may be supplied by either or both of the pulmonary or systemic arterial system, and drain to pulmonary or systemic veins.

Causes: No definite known cause although there is interest in the role of some genes such as fatty acid binding protein-7 in the pathogenesis of CPAMs. The overlap and occasional coexistence of these cystic lesions suggest a possible single common pathologic mechanism


Thank you A/Prof Adam Jaffe. Please download this [Excel Aide memoire file](#) to help you to keep track of your reported cases
 Please type the number of cases you have seen then click on the "Submit" button. For example if you have seen 2 cases of Loeffler syndrome and one case of ChILD then type 2 in the Loeffler syndrome box and 1 in the ChILD box. leave other boxes blank.

ENTER DETAILS OF NEWLY PRESENTING CASES BELOW	Number seen
Congenital malformations	
Trachea	<input type="text"/>
Tracheo-oesophageal fistula	<input type="text"/>
Diaphragmatic hernia	<input type="text"/>
Cystic lung lesions	<input type="text"/>
Lobar emphysema	<input type="text"/>
Pleural/Pulmonary lymphangiectasia	<input type="text"/>
Grown up (>16) chronic neonatal lung disease	<input type="text"/>
Lymphangiomyomatosis	<input type="text"/>
Alveolar haemorrhage syndrome	
Goodpasture syndrome	<input type="text"/>
Idiopathic pulmonary hemosiderosis	<input type="text"/>
Hered hemor telang (HHT) / (OWR) syndrome	<input type="text"/>
Langerhans cell histiocytosis	<input type="text"/>

Patient Discussion Forum



ARNOLD
Australasian Registry Network
for Orphan Lung Disease



simple machines for

USER INFO

Welcome, **Guest**. Please [login](#) or [register](#).
Did you miss your [activation email](#)?
February 07, 2010, 10:19:12 PM

Forever


Login with username, password and session length

NEWS BOX

KEY STATS


1 Posts in 1 Topics by 5 Members
Latest Member: [pinkhaus27](#)

Search: [Advanced search](#)

 **arnold**

Members: 5 • Posts: 1 • To
Please welcome [pinkhaus27](#) our newest m

Patients and Parents



General Discussion
Any topics related to Orphan Lung Disease

1 Posts in
1 Topics

Last post on February 22, 2009, 05:1
AM
in [Welcome to the ARNOLD bu...](#) by
ARNOLD administrator

Links

Links

Site	Description
Australian Lung Foundation	
Australian Thoracic Society of Australia and New Zealand	
Adult Interstitial Lung Disease Information	
Infant Interstitial Lung Disease Information	
BreatheOn	Uk National charity supporting all young people up to the age of 21 years who are either ventilated or technology dependent for over three months.
British Thoracic Society	
Orphanet	

[Primary Ciliary Dyskinesia Family Support Group](#)

[European Organisation for Rare Diseases](#)

[British Paediatric Respiratory Society](#)

[Contact a Family](#) For families with disabled children

[British Paediatric Surveillance Unit](#) (RCPCH)

[National Organisation for Rare Disorders](#)

[chILD Foundation](#) Parental support group in USA for Interstitial Lung Disease

[Children Interstitial Lung Disease Yahoo Group](#) Chat room for parents with Interstitial Lung Disease

[Little Leakers](#) USA site for children with Lymphangiectasia

Rebecca's Solution



Orphan Lung Diseases... ...A Soft Place To Land...

Home
About Me
Orphan_Lung_Diseases
Follicular_Bronchiolitis
The_AutoImmune_Connection
Chronic_Illness
Living_with_a_Disability
Invisible_Disabilities
Support_for_Carers
Hopes_and_Aspirations
Reference_Page
Acknowledgements_and_Links
Contact_us
A_Word_of_Thanks

I believe that everyone needs a 'Soft Place to Land'... A place where they feel safe, secure, understood and encouraged.

Life throws many challenges our way even when we are fit, healthy and without illness. For those of us who have an illness or disability-we know all too well that life will continue to throw the usual difficulties our way, but over and above these difficulties, we are also faced with a whole range of challenges that are unique to having an illness or disability. We face our own

What difference can we really make?

- Sanjad-Sakati Syndrome
 - Autosomal recessive
 - Congenital Hypoparathyroidism
 - Short Stature
 - Mild mental retardation
 - Dysmorphic features

- AD , 15 Yrs
- SSS
- Delayed diagnosis
- 2 sisters with same syndrome
(1 Died at 6 months of age, other 5 yrs old)
- Fathers Education:
 - Elementary school
 - Office clerk
- Father frustrated with late diagnosis and lack of knowledge by doctors



Sanjad-Sakati Website

http://www.sanjadsakati.co.uk

الجمعية الإلكترونية
لإتلازمة
ساجد سكاتي
www.sanjadsakati.org

الموقع الرسمي لمتلازمة ساجد سكاتي

الرئيسية

عن الجمعية

ساجد سكاتي

أخبار الجمعية

خدمات الجمعية

استشارات طبية

المسجل الوطني

المنتدى

ألبوم الصور

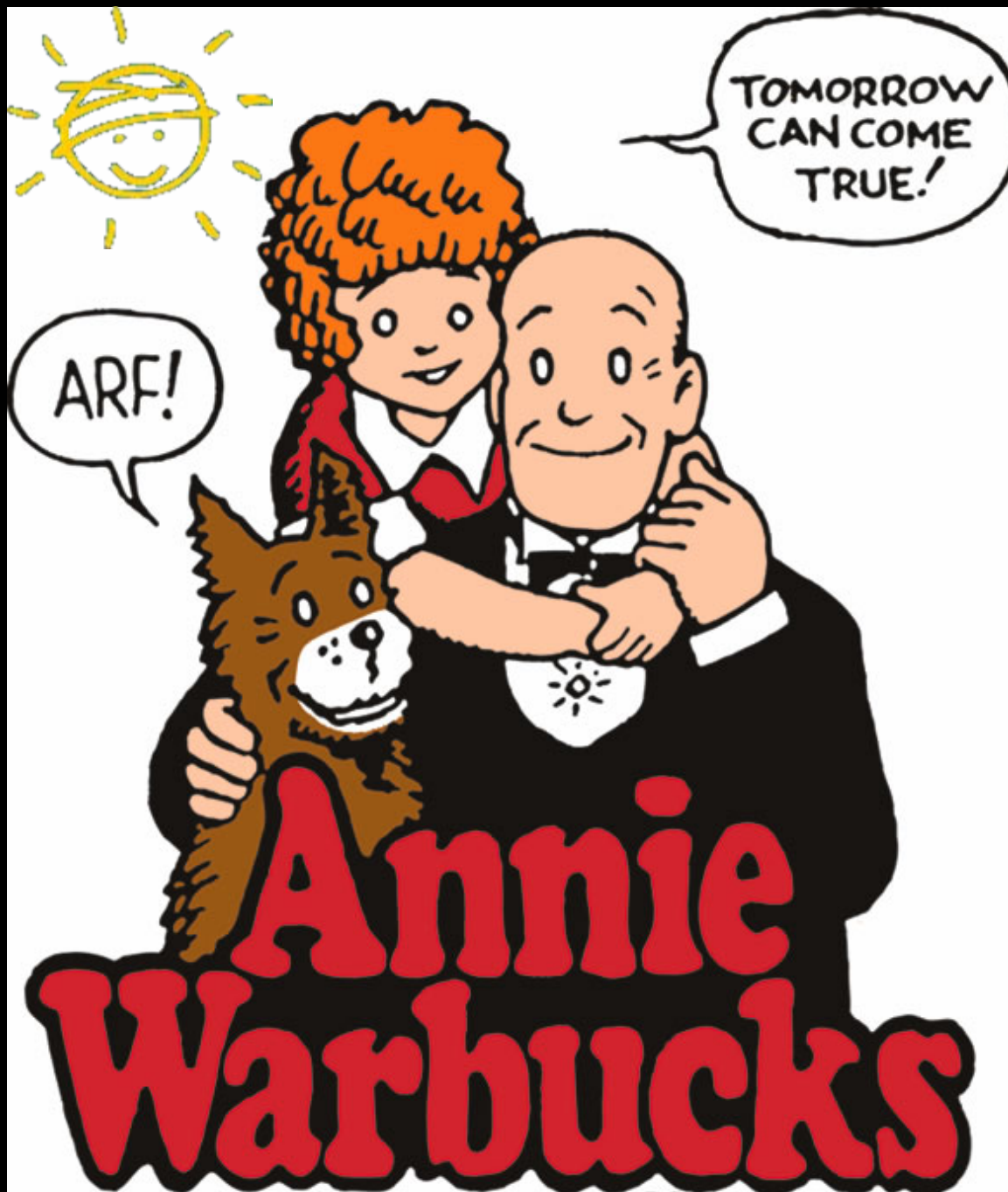
العدد لله وحده والصلاة والسلام على من لا نبي بعده محمد ابن عبدالله وعلى آله وصحبه وبعد النفس البشرية من معجزات الله في خلقه سبحانه وتعالى ومن نعم الله علينا الجليلة أن خلقنا أنبياء وسفر لنا العقل والحواس والفكر والعقل السليم في الجسم السليم لا يزال مقيوم الإعاقات وأوعيا وأبوابها ونصحتها ومشاكلها النفسية والسلوية والتأهيلية غير واضح لبعض فئات المجتمع كما في هذا المرض والأشهر العربية لديها الكثير من المعوقات العنصرية والاجتماعية التي تقلل برعايتها للطفل ذوي الإعاقة من الإحالة لهم لخدمة اجتماعية ومهنية والتمهيد لهم إمكانية العمل على تقليل حجمها ورفع قدراتها وإلا فمن شأنها النفسية الاجتماعية الاقتصادية والتعليمية على المعاناة التي قد تلحقها من عدم حصولها على الخدمات المناسبة.

2 100% Internet

- Started an Association
- Printing and translating information from internet for others
- Planning a national medical and public conference
- Creating a database

Summary

- Orphan respiratory diseases are a significant health burden
- Lack of information for clinicians and patients frustrating
- Increasing interest in orphan lung diseases
- Collaboration essential
- Consumer pressure push rare lung diseases up political agenda
 - Empowering patients and their families
- Politicians need to be made aware
 - Need for coordinated National Plan
- Tomorrow....?



Thanks to

- Dr Yvonne Zurynski
 - Australian Paediatric Surveillance Unit
- Dr Abdullah Yousef
 - Respiratory Fellow
 - Sydney Children's Hospital, Randwick
 - Saudi Arabia