Clinical problems in rare interstitial lung diseases

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Epidemiology of childhood interstitial lung disease (chILD) in France : The RespiRare cohort

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Introduction

Childhood interstitial lung disease (chILD) is a heterogeneous group of rare lung diseases that share common radiological features of interstitial signs on chest X-ray or chest computed tomography (CT)-scan.

ChILD can present throughout life, from birth to adolescence, **with important variations in severity from pauci-symptomatic diseases to end-stage respiratory failure leading to pulmonary transplantation or death**. ChILD classification usually distinguish two age groups: below 2 years and 2 to 18 years.

Limited number of patient series with chILD have been reported worldwide by single centers or large networks (Table 1).

The present study aims at describing the epidemiology of chILD in France in terms of prevalence, incidence, respective frequencies of the different etiologies of ILD and age of onset.

Table 1: chILD series reported to date

Country / Area	Patients (n)	Period of time (years)	References
United Kingdom	46	3	*Dinwiddie et al. 2002*
and Ireland	46	1	*Laverty et al. 2008*
Denmark	884	10	*Kornum et al. 2008*
Germany	56	1	*Griese et al. 2009*
France	205	4	*Nathan et al. 2012*
Australia and New	71	5	*Casamento et al. 2016*
Zealand	115	10	*Saddi et al. 2017*
USA	93	18	*Soaers et al. 2013*
	256	1	*Young et al. 2018*
China	133	5	*Tang et al. 2020*
Europe and above	575		*Griese et al. 2017*
Spain	381	2	*Torrent-Vernetta et al. 2022*

Methods

Type of study
- Observational
- Retrospective (2000 - 2022)
- Prospective (Feb 2022-Feb 2023)
- National, multicentric
- Legal issue: ethics committee gave their authorization

Inclusion criteria
- Patients with ILD defined following the ATS Clinical Practice Guideline
- Born between 2000 to 2022 and aged under 18-year-old at diagnosis

Exclusion criteria
- Bronchopulmonary dysplasia,
- No available data

Data collection
- e-Respirare
- Multidisciplinary team meetings (MDT) for chILD
- Direct emails to the centres

Localisation of the study
All the RespiRare centres in France

Results

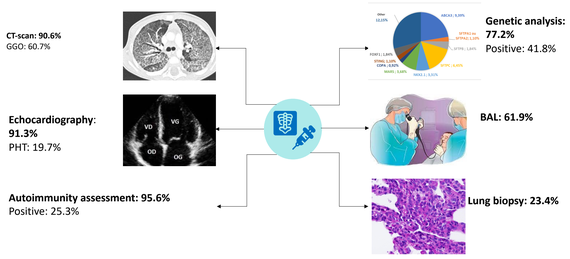
A total of 790 patients were included

Median age at diagnosis: 3 m
Premature birth :17.7%
Sex ratio: 1
Family history of ILD: 16.9%

The prevalence of chILD in 2022 was 44.04/million of children

The incidence of chILD in 1 year was 4.44/million of children

Investigations performed:



chILD etiologies

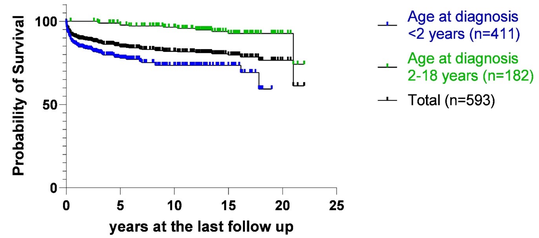
Below 2 years, the main ILD etiologies were surfactant metabolism disorders and proteinosis alveolar (23.7%) and NEHI (11.8%).

Between 2 and 18 years, the main etiological diagnoses were alveolar hemorrhage and pulmonary hemosiderosis (12.2%), connective tissue diseases (11.4%), hypersensitivity pneumonitis (8.8%), sarcoidosis (8.8%) and vascularitis (7%).

Results

Evolution

At follow up: Deceased patients: 17.7%
5 years survival (Figure 1):
- For patients diagnosed < 2 years: 57.3%
- For patients diagnosed > 2 years: 86%



Kaplan Meier survival according to the age at diagnosis.

Discussion and conclusion

This large epidemiological study of chILD confirms a **higher incidence and prevalence than previously reported.** Our findings are similar to the 2022 Spanish study.

The next challenge will be to take advantage of well-organized international networks such as the CRC chILD and the ERN-lung to develop systematic cohorts and registers – at least for some chILD etiologies - to develop and test new therapeutic options and to prepare new guidelines for the management and transition of care of these patients.

RespiRare Centre de référence des maladies respiratoires rares

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