

| Demographics | |
|---|---|
| Sample number: _____ | Family number: _____ |
| Patient surname: _____ | Patient forenames: _____ |
| Date of birth: _____ | Gender: _____ |
| Address: _____ _____ | State: _____ Postcode: _____ |
| Deceased: <input type="checkbox"/> Yes <input type="checkbox"/> No | Date of death: _____ Cause of death: _____ |
| Date of first medical presentation: _____ | Date of presumed diagnosis based on biochemical/histological studies performed: _____ |
| Date of first specialist clinic consult: _____ | Name of specialist service: _____ |
| Final diagnosis: _____ | Age at diagnosis: _____ |
| Weight at diagnosis kg (percentile): _____ | Height at diagnosis cm (percentile): _____ |
| Ethnicity: <input type="checkbox"/> Aboriginal /Torres Strait Islander <input type="checkbox"/> Australian/ New Zealander <input type="checkbox"/> North African/Middle Eastern <input type="checkbox"/> Maori/Pacific Islander <input type="checkbox"/> Asian <input type="checkbox"/> Sub-Saharan African <input type="checkbox"/> Other Oceanian <input type="checkbox"/> People of the Americas <input type="checkbox"/> Sephardic Jew <input type="checkbox"/> European (Non-Finnish) <input type="checkbox"/> European (Finnish) <input type="checkbox"/> Ashkenazi Jew <input type="checkbox"/> Declined to specify/unknown | |
| Family history & pedigree – for families with multiple affected individuals, identify index case. Attach pedigree if available. | |
| | |
| History of childhood interstitial lung disease: <input type="checkbox"/> Maternal <input type="checkbox"/> Paternal | |
| Maternal ethnicity: _____ | Paternal ethnicity: _____ |
| Maternal age: _____ | Likely to have more children: <input type="checkbox"/> Yes <input type="checkbox"/> No |
| Currently pregnant | <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown |
| Consanguinity | <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown |
| Siblings affected by similar disease | <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown |

| | | | |
|---|---|--|----------------------------------|
| Other affected family members | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Specify: _____ | | | |
| Patient history | | | |
| Gestation (wk): _____ | Birth Weight (g): _____ | | |
| Respiratory distress at birth: <input type="checkbox"/> Yes <input type="checkbox"/> No | Delivery: <input type="checkbox"/> SVD <input type="checkbox"/> Caesarean | | |
| Age at disease onset: _____ | Age at first hospital admission: _____ | | |
| Required intubation? <input type="checkbox"/> Yes. <input type="checkbox"/> No | Required surfactant? <input type="checkbox"/> Yes <input type="checkbox"/> No | | |
| Duration of symptoms when first seen | _____ | | |
| Other syndromic features/ phenotypes | _____ | | |
| Other concomitant disease | _____ | | |
| Autoimmune disease | <input type="checkbox"/> Rheumatoid arthritis <input type="checkbox"/> Systemic Lupus Erythematosus <input type="checkbox"/> Unknown <input type="checkbox"/> Other: _____ | | |
| Atopy | <input type="checkbox"/> Asthma <input type="checkbox"/> Eczema <input type="checkbox"/> Hay fever | | |
| Other respiratory disease: | _____ | | |
| Previous serious lower respiratory tract infection | <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Unknown |
| Differential diagnoses (please give multiple diagnoses as will help the laboratory identify the disease gene) | | | |
| | | | |
| Suspected diagnosis | | | |
| <input type="checkbox"/> child | <input type="checkbox"/> Diffuse Lung Disease | | |
| <input type="checkbox"/> Pulmonary Fibrosis | <input type="checkbox"/> Idiopathic Pulmonary Haemosiderosis | | |
| <input type="checkbox"/> Pulmonary Alveolar Proteinosis | <input type="checkbox"/> Pulmonary Hypertension | | |
| <input type="checkbox"/> Lung Development Disorder | <input type="checkbox"/> Cystic Lung Disease | | |
| Age of onset of presenting symptoms | | | |
| <input type="checkbox"/> Prenatal | <input type="checkbox"/> Childhood (>2 to 18 years) | | |
| <input type="checkbox"/> Neonatal (Birth to 28 days) | <input type="checkbox"/> Adult (>18 years) | | |
| <input type="checkbox"/> Infantile (> 28 days month to 2 year) | | | |
| Evolution of symptoms | | | |
| <input type="checkbox"/> Rapidly progressive (<4 weeks) | <input type="checkbox"/> Slowly progressive (>4 weeks) | | |
| <input type="checkbox"/> Non-progressive | <input type="checkbox"/> Acute-episodic (relapsing-remitting) | | |
| Symptoms at onset | | | |
| <input type="checkbox"/> Cough | <input type="checkbox"/> Tachypnoea | <input type="checkbox"/> Exertional dyspnoea | |
| <input type="checkbox"/> Dyspnoea at rest | <input type="checkbox"/> Fever | <input type="checkbox"/> Failure to thrive | |
| <input type="checkbox"/> Others: _____ | | | |
| Symptoms throughout treatment | | | |

| | | |
|--|---|--|
| <input type="checkbox"/> Cough | <input type="checkbox"/> Tachypnoea | <input type="checkbox"/> Exertional dyspnoea |
| <input type="checkbox"/> Dyspnoea at rest | <input type="checkbox"/> Fever | <input type="checkbox"/> Failure to thrive |
| <input type="checkbox"/> Others: _____ | | |
| Physical examination | | |
| <input type="checkbox"/> Cyanosis | <input type="checkbox"/> Crackles | <input type="checkbox"/> Clubbing |
| <input type="checkbox"/> Wheeze | <input type="checkbox"/> Failure to thrive | <input type="checkbox"/> Tachypnoea |
| <input type="checkbox"/> Others: _____ | | |
| Minor neural dysfunction | <input type="checkbox"/> Yes | <input type="checkbox"/> No |
| Fine manipulative disability | <input type="checkbox"/> Yes | <input type="checkbox"/> No |
| Dyscoordination | <input type="checkbox"/> Yes | <input type="checkbox"/> No |
| Excessive associated movements | <input type="checkbox"/> Yes | <input type="checkbox"/> No |
| Investigations at Diagnosis | | |
| Chest imaging | | |
| Chest films | Interstitial infiltrates | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| | Alveolar infiltrates | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| | Other: | _____ |
| CT scans | Reticular, nodular infiltrates | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| | Ground glass pattern | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| | Honeycomb pattern | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| | Mosaic pattern | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| | Other: | _____ |
| Was the CT scan performed with controlled ventilation? | <input type="checkbox"/> Yes | <input type="checkbox"/> No |
| Did the CT have expiratory views? | <input type="checkbox"/> Yes | <input type="checkbox"/> No |
| Pulmonary function | | |
| Done? <input type="checkbox"/> Yes <input type="checkbox"/> No (go to next question) | | |
| FEV1 (L) _____ (%) | FVC (L) _____ (%) | |
| FRC (L) _____ (%) | TLC (L) _____ (%) | |
| RV(L) _____ (%) | RV/TLC ratio _____ (%) | |
| CO transfer _____ (%) | FEV1 /FVC ratio _____ (%) | |
| Infant PFT | | |
| Done? <input type="checkbox"/> Yes <input type="checkbox"/> No (go to next question) | | |
| What type (e.g. raised volume, lung clearance index)? | | |
| Result: | | |
| <input type="checkbox"/> Normal <input type="checkbox"/> Abnormal (please attach result) | | |
| Gas exchange – in room air | | |
| Done? <input type="checkbox"/> Yes <input type="checkbox"/> No (go to next question) | | |
| SaO ₂ _____ (%) | PaO ₂ (mmHg): _____ | PaCO ₂ (mmHg): _____ pH: _____ |
| Sleep study – in air or in O₂ _____ L/Min | | |
| Done? <input type="checkbox"/> Yes <input type="checkbox"/> No (go to next question) | | |
| Mean O ₂ saturations _____ (%) | Lowest O ₂ saturations _____ (%) | |
| Exercise testing | | |
| Done? <input type="checkbox"/> Yes <input type="checkbox"/> No (go to next question) | | |
| What type? | | |

| | | |
|--|---|--|
| Result: | | |
| <input type="checkbox"/> Normal <input type="checkbox"/> Abnormal (please attach result) | | |
| Echocardiogram | | |
| Done? <input type="checkbox"/> Yes <input type="checkbox"/> No (go to next question) | | |
| Result: | | |
| <input type="checkbox"/> Normal <input type="checkbox"/> Abnormal (please attach result) | | |
| Pulmonary hypertension | | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| Bronchoalveolar lavage | | |
| Done? <input type="checkbox"/> Yes <input type="checkbox"/> No (go to next question) | | |
| Macroscopic comment _____ | | Cell number 10 ³ /ml _____ |
| Macrophages _____ (%) | Lymphocytes _____ (%) | CD4+/CD8+ _____ (%) |
| Neutrophils _____ (%) | Eosinophils _____ (%) | CD1a+ _____ (%) |
| PAS staining (if positive %) _____ (%) | Haemosiderin-staining- (if positive %) _____ (%) | Lipid-staining (if positive %) _____ (%) |
| Microbiology at diagnosis | | |
| Done? <input type="checkbox"/> Yes <input type="checkbox"/> No (go to next question) | | |
| Pathogen(s) identified: | | |
| <input type="checkbox"/> Adenovirus | <input type="checkbox"/> Pneumocystis | <input type="checkbox"/> Aspergillus |
| <input type="checkbox"/> Cytomegalovirus | <input type="checkbox"/> Epstein Barr Virus | <input type="checkbox"/> Mycoplasma |
| <input type="checkbox"/> Influenza virus | <input type="checkbox"/> HIV | <input type="checkbox"/> Mycobacteria + TB |
| <input type="checkbox"/> Parainfluenza virus | <input type="checkbox"/> Chlamydia | <input type="checkbox"/> Legionella |
| <input type="checkbox"/> Respiratory syncytial virus | <input type="checkbox"/> Other: | |
| Methods (specify): | | |
| Cultures <input type="checkbox"/> Yes <input type="checkbox"/> No | Immunofluorescence <input type="checkbox"/> Yes <input type="checkbox"/> No | |
| Other: | | |
| Respiratory screening (serology) | | |
| Done? <input type="checkbox"/> Yes <input type="checkbox"/> No (go to next question) | | |
| Viral antibody | | |
| Adenovirus: <input type="checkbox"/> positive <input type="checkbox"/> not done | RSV: <input type="checkbox"/> positive <input type="checkbox"/> not done | |
| Herpes Simplex: <input type="checkbox"/> positive <input type="checkbox"/> not done | Influenza B: <input type="checkbox"/> positive <input type="checkbox"/> not done | |
| Influenza A: <input type="checkbox"/> positive <input type="checkbox"/> not done | Varicella Zoster: <input type="checkbox"/> positive <input type="checkbox"/> not done | |
| Mumps: <input type="checkbox"/> positive <input type="checkbox"/> not done | Parainfluenza: <input type="checkbox"/> positive <input type="checkbox"/> not done | |
| Epstein Barr: <input type="checkbox"/> positive <input type="checkbox"/> not done | | |
| Other respiratory pathogens (<i>specify if abnormal</i>) | | |
| Pneumocystis: <input type="checkbox"/> positive <input type="checkbox"/> not done | Aspergillus: <input type="checkbox"/> positive <input type="checkbox"/> not done | |
| Chlamydia: <input type="checkbox"/> positive <input type="checkbox"/> not done | Legionella: <input type="checkbox"/> positive <input type="checkbox"/> not done | |
| Mycoplasma IgG: <input type="checkbox"/> positive <input type="checkbox"/> not done | TB: <input type="checkbox"/> positive <input type="checkbox"/> not done | |
| Mycoplasma IgM: <input type="checkbox"/> positive <input type="checkbox"/> not done | | |
| Immunology (<i>specify if abnormal</i>) | | |
| Immunoglobulin | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ | |
| T and B subsets | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ | |
| Response to immunisations | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ | |
| Hypersensitivity screening | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ | |
| Autoantibodies | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ | |
| Complement | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ | |
| Other | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ | |

| Haematology (specify if abnormal) | |
|---|---|
| Full blood count | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ |
| White cell count | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ |
| Platelets | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ |
| Haemoglobin | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ |
| Cardiology (please attach result if abnormal) | |
| Done? <input type="checkbox"/> Yes <input type="checkbox"/> No (go to next question) | |
| ECG, echocardiogram: | _____ |
| Others (specify if abnormal) | |
| ACE | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ |
| Rheumatoid factor | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ |
| Autoantibodies (serum) | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ |
| Ciliary study | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ |
| Thyroid function test | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ |
| Others | Done? <input type="checkbox"/> No <input type="checkbox"/> Yes: _____ |
| Gastro-oesophageal reflux investigations | |
| Done? <input type="checkbox"/> Yes <input type="checkbox"/> No (go to next question) | |
| What type? (e.g. pH/impedance/barium/endoscopy) | |
| _____ | |
| Result (please attach) | |
| _____ | |
| Lung biopsy | |
| Done? <input type="checkbox"/> Yes <input type="checkbox"/> No (go to next question) | |
| How? | <input type="checkbox"/> Open |
| | <input type="checkbox"/> Video-assisted thorascopic surgery |
| | <input type="checkbox"/> Transbronchial |
| Age at biopsy: | _____ |
| Date of biopsy: | _____ |
| Histology performed at local centre? | <input type="checkbox"/> Yes <input type="checkbox"/> No (where?): _____ |
| Second opinion sought? | <input type="checkbox"/> No <input type="checkbox"/> Yes (where?): _____ |
| Result (please insert de-identified histopathology formal report) | |
| _____ | |
| Genetics | |
| Done? <input type="checkbox"/> No (go to next question) <input type="checkbox"/> Yes, where? | <input type="checkbox"/> Children's Hospital at Westmead <input type="checkbox"/> Other: |
| Chromosomal abnormality present? | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| Results (please attach) | |
| _____ | |
| Treatment | |
| Respiratory support | |
| Maximum oxygen treatment required | _____ L/Min |
| Feeding | |
| Nutritional support (oral or enteral) | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| Gastrostomy fed | <input type="checkbox"/> Yes <input type="checkbox"/> No |
| Initial drug treatment: eg Steroids, Hydroxychloroquine, Azithromycin, Cytotoxic drug, others | |
| _____ | |

| Drug name | Date started | Daily dosage | Date finished | Response [#] | |
|-----------|--------------|--------------|---------------|--------------------------|--------------------------|
| | | | | Yes | No |
| | | | | <input type="checkbox"/> | <input type="checkbox"/> |
| | | | | <input type="checkbox"/> | <input type="checkbox"/> |
| | | | | <input type="checkbox"/> | <input type="checkbox"/> |
| | | | | <input type="checkbox"/> | <input type="checkbox"/> |
| | | | | <input type="checkbox"/> | <input type="checkbox"/> |
| | | | | <input type="checkbox"/> | <input type="checkbox"/> |

#Improvement in physiological outcomes considered to be a treatment response

| | Possible response (%) | Response (%) | Best Response (%) |
|---|-----------------------|--------------|-------------------|
| Heart rate | 10 | 20 | - |
| Respiratory rate | 5 | 10 | 20 |
| SpO ₂ | 5 | 10 | - |
| Loss of need for supplemental oxygen | - | - | Yes |
| Loss of need for mechanical ventilation | - | - | Yes |

Conversely, a deterioration of this size would be considered a significant decline. SpO₂, arterial oxygen saturation.

Bush A, Cunningham S, de Blic J, et al. *Thorax* 2015; **70**:1078–1084

Follow up and latest outcome

Good*: improvement in tachypnoea and respiratory difficulty, return of weight gain and growth towards normal, improvement in exercise tolerance, lung function and oxygen saturation at rest.

Poor**: failure to improve in the manner described above despite treatment.

| | |
|-----------------------------|--|
| Clinical progress | <input type="checkbox"/> Good* <input type="checkbox"/> Poor** <input type="checkbox"/> Transplant <input type="checkbox"/> Died |
| Oxygen saturation | _____%. _____ Insp O ₂ . _____ n/d |
| Lung function | FEV1 _____% |
| | FVC _____% |
| Radiological changes | <input type="checkbox"/> Chest X-Ray <input type="checkbox"/> CT |
| | <input type="checkbox"/> Improved <input type="checkbox"/> Deterioration |
| | <input type="checkbox"/> No change <input type="checkbox"/> Not done |
| Any other findings/comments | |

Hospital discharge codes on initial presentation (International Classification of Diseases codes):

| | | |
|-----------------------|----------------|------------------|
| Admission date: _____ | Primary: _____ | Secondary: _____ |
|-----------------------|----------------|------------------|

Hospital discharge codes on subsequent admissions:

| | | |
|-----------------------|----------------|------------------|
| Admission date: _____ | Primary: _____ | Secondary: _____ |
| Admission date: _____ | Primary: _____ | Secondary: _____ |
| Admission date: _____ | Primary: _____ | Secondary: _____ |
| Admission date: _____ | Primary: _____ | Secondary: _____ |
| Admission date: _____ | Primary: _____ | Secondary: _____ |
| Admission date: _____ | Primary: _____ | Secondary: _____ |
| Admission date: _____ | Primary: _____ | Secondary: _____ |

Classification of ChILD (Deutsch et al. Am J Resp Crit Care Med 2007:176;1120- 1128)

| DISORDERS MORE PREVALENT IN INFANCY | DISORDERS LESS PREVALENT IN INFANCY |
|---|--|
| <p>Diffuse developmental disorders of the lung: Acinar dysplasia Congenital alveolar dysplasia Alveolar capillary dysplasia with misalignment of pulmonary veins</p> | <p>Disorders related to systemic disease processes: Immune mediated/collagen vascular disorders Storage disease Sarcoidosis Langerhans cell histiocytosis Malignant infiltrates</p> |
| <p>Lung growth abnormalities reflecting deficient alveolarisation: Pulmonary hypoplasia Chronic neonatal lung disease Related to chromosomal disorders Related to congenital heart disease</p> | <p>Disorders of the normal host: Related to infections Related to environmental agents hypersensitivity pneumonitis toxic inhalation Aspiration syndromes Eosinophilic pneumonia</p> |
| <p>Specific conditions of undefined cause: Neuroendocrine cell hyperplasia of infancy Pulmonary interstitial glycogenosis</p> | <p>Disorders masquerading as ILD: Arterial hypertensive vasculopathy Congestive changes related to cardiac dysfunction Veno-occlusive disease Lymphatic disorders</p> |
| <p>Inherited surfactant disorders: Surfactant protein B mutation Surfactant protein C mutation <i>ABCA3</i> mutations Histology consistent with the surfactant dysfunction without a yet recognised genetic aetiology</p> <ul style="list-style-type: none"> • pulmonary alveolar proteinosis • chronic pneumonitis of infancy • desquamative interstitial pneumonitis • non-specific interstitial pneumonia | |