International Platform for Childhood Interstitial Lung Diseases

European chILD Register and Biobank

chILD-EU

COOPERATION

Living with chILD

An Information Booklet for Parents and Relatives of affected Children

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PREFACE

Dear Parents, Family and Relatives,

If you are holding this booklet in your hands, then your child has already been diagnosed with an interstitial lung disease (**chILD**) or it has been raised as a potential diagnosis.

"**chILD**" is an acronym for a multitude of differing chronic diseases, all sharing in common the symptom of dyspnoea (shortness of breath) and often also having hypoxia (decreased oxygen uptake via the lungs). The acronym has been derived from the English name for this group of diseases, Children's Interstitial Lung Diseases – **chILD**.

In view of the rarity of these diseases in children, there are only a very limited number of doctors significantly familiar with them, especially in terms of diagnosing and treating them. A search of the internet for further information unfortunately is often more troubling than beneficial. Frequently people develop anxiety, fear or melancholy when trying to deal with these chronic diseases, because of the limited knowledge about them and their causes. This is often exacerbated by poor or limited information being provided by their doctors, regarding investigations, treatments and strategies to conquer their disease. Additionally, this can contribute to feelings of being helpless and isolated.

With aid from the European Union, an international expert panel has been formed, that has set the goal to correct this situation. An additional goal is to place the needs of the children afflicted with these diseases and their families first. This booklet is only a single building block in this project. Of course, it cannot replace the need for talking with your child's treating doctor. Rather, it is intended to provide supplementary information regarding the condition of your child.

It is not necessary to read everything in a single sitting, as each chapter has been inspired by frequently asked questions from parents and children. Maybe you will find in this handbook the answer to questions you have previously asked, but have not received an adequate answer for.

Your opinion is very important to us! We would be very grateful and appreciative for feedback regarding this booklet; whether you liked it, think something is missing or is written in a confusing manner.

We would like to give our thanks to all the children and their parents, who have helped us develop this handbook and by allowing us to tell their stories. Special thanks go out to the mum of one child, Mrs. Judith Eisenbach, who has given us invaluable support in writing this booklet. In the hope that this handbook is a little helpful for you and with the best wishes for you, your children and your whole family.

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What is the chILD-EU Project?

ChILD-EU is a project funded through the European Union, which was started in December 2012 and will be financed over a period of 42 months (**http://www.klinikum.uni-muenchen.de/Child-EU/en/index.html**). As explained in the preface, the term is an acronym derived from the English abbreviation for a group of lung diseases, **ch**ildren's **i**nterstitial **l**ung **d**isease, forming the word "child".

The goal of this project is to gain more knowledge about the causes of the diseases, their progress over time, available diagnostic techniques and therapeutic options, through a systematic review of information available about these rare lung diseases. Additionally, the treatment of children in Europe with these diseases will be improved and unified through the publication of diagnostic and therapeutic guidelines.

Another very important goal is to develop an information and education programme for children with the disease and their relatives. This is in an effort to make it easier for them to deal with the chronic illness. In **chapter 12** there is further information provided about this project and research into chILD.

CHAPTER 1: THE DEVELOPMENT AND FUNCTION OF THE AIRWAYS

How do the airways develop?

The completed development of the human body from a fertilised egg and the function of each of its organs (such as the lungs) are incredibly complex. Lack of understanding of these processes can make it even more difficult for patients and their relatives to understand the actual cause or causes of their medical illness. This can be made even more difficult by the fact that doctors often use words or terms when explaining things that they themselves may not fully understand. Further, the flood of information provided during each short discussion with your health provider, can often be too much to subsequently remember or fully grasp. Which often leads to questions such as: "What have I been told (again)?", "What problem do I actually have?", "Where did it come from?" and "What is coming next?".

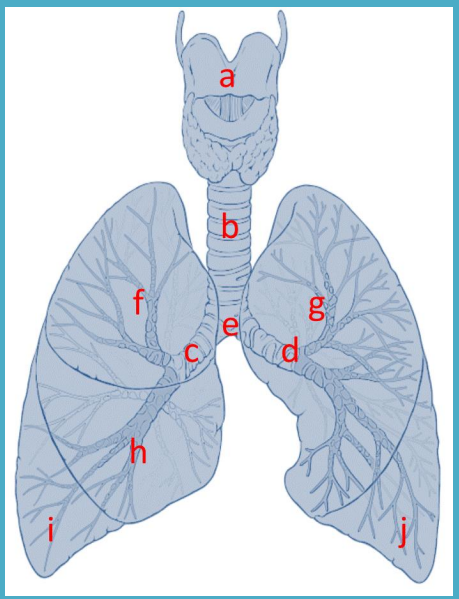
To better understand a disease and its origin, it is helpful to know a bit more about the development and function of the affected organs. That is why this chapter is dedicated to the development and function of the airways. Additionally, it will also explain what fundamental changes occur as a result of having **chILD** (interstitial lung diseases of childhood).

The Lung

The lung is a paired organ in the chest (thorax), whose primary function is to enable breathing. It consists of two "wings" on each side of the chest, a left and a right, which are moderately different in design. The right lung wing consists of three lobes (an upper, middle and lower lobe), while the left has only two lobes (an upper and lower lobe) (***see* Figure 1**). The structures that guide air into the alveoli (the terminal sack-like compartments of each lobe) are called the airways. The airways are divided into an upper and lower part. The upper airway includes the nose, the mouth and the throat. The larynx (voice box) is the point of transition to the lower airways, which is made up of the trachea, bronchi and bronchioles.

Figure 1: Schematic picture of the Airways and Lungs:

a Voice Box (or Larynx)
b Windpipe (or Trachea)
c Right Main Bronchus
d Left Main Bronchus
e Carina
f Right Upper Lobe
g Left Upper Lobe
h Right Middle Lobe
i Right Lower Lobe
j Left Lower Lobe



Trachea and Bronchi

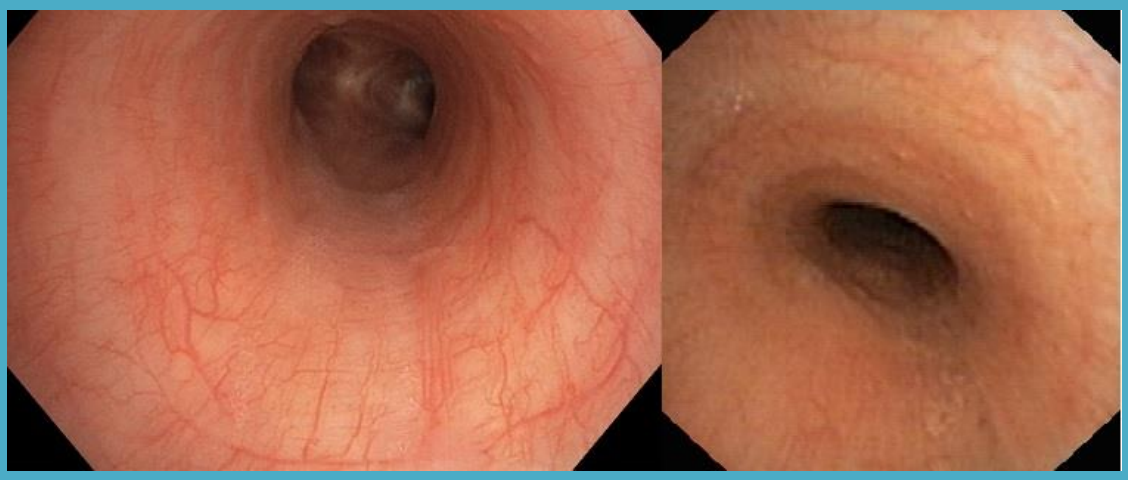
The trachea (windpipe) consists of horseshoe shaped "rings" of cartilage that are connected with each other by tough, fibrous ribbons of tissue (ligaments). These cartilage rings provide stability to the trachea and hold it open (***see* Figure 2**). There are diseases where the cartilage rings are very soft, hence causing the wind pipe to partially or completely collapse when breathing. This problem is called *Tracheomalacia* (***see* Figure 2**). Whereas those children that have only mild forms of this condition only become apparent when they demonstrate an abnormally roaring cough, those with severe forms present earlier with severe breathing difficulties or shortness of breath (dyspnoea). Another problem, known as *Tracheal Stenosis*, occurs when the rings are too small or form a complete circle, which results in narrowing of the airway. The severity of this illness is directly related to the extent and severity of the airway narrowing. If present, tracheomalacia may be made worse by lung disease in ChILD. Tracheal stenosis is not normally present in ChILD.

At the lower end the trachea splits into the right and left main bronchi (***see* Figures 1 and 2**). The right main bronchus splits into the right upper, middle and lower lobe bronchi, whilst the left main bronchus splits only into the left upper and lower lobe bronchi. These lobar bronchi then subsequently divide into smaller and then smaller bronchi, much like one can imagine branching on a tree from its main trunk out to the leaves. The smallest bronchi are called bronchioles. These branch even further and terminate at the alveoli (***see* Figure 3**).

Figure 2: Bronchoscopy of the trachea

Left Picture: a photo of the inside of a normal healthy trachea (windpipe), taken during a bronchoscopy. The epithelium (surface) shows no irritation (inflammation) and the airway is being held open by cartilage rings (visible along the top of the pipe). At the far end you can see the division into the right and left main bronchi the carina.

Right Picture: A photo of a partially collapsed soft trachea with ovoid narrowing, seen with tracheomalacia.

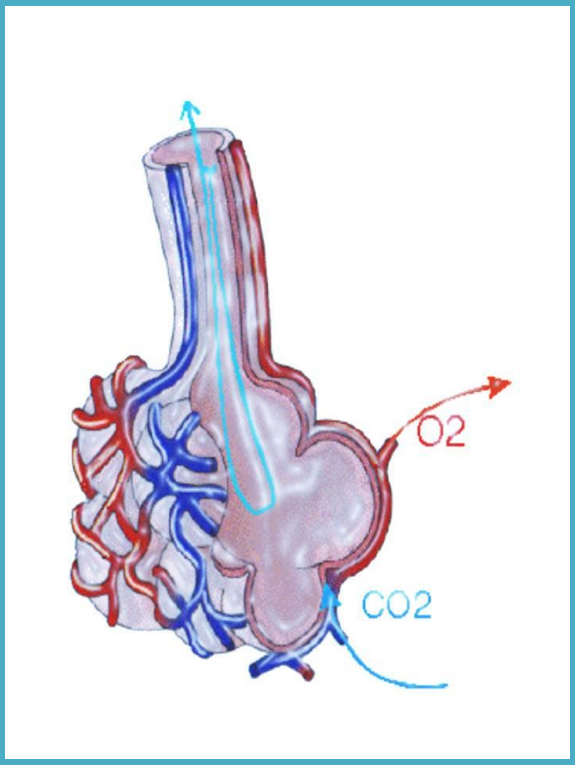


A

The lung vesicles (alveoli) are grape-shaped sacks at the ends of the tiniest pipes of the airways (the bronchioles). The lung of a new-born baby has approximately 100 million alveoli, as opposed to an adult with 300 million. A network of our tiniest blood vessels (capillaries) fuse onto the surface of each of these very thinly walled alveoli (***see* Figure 3**). This enables an efficient and effective way for the uptake of oxygen from the air into the blood and also the transfer of waste carbon dioxide from the blood back into the air. The walls of each alveolus are lined with two highly specialised cells, Type 1 and Type 2 Pneumocytes. The main role of Type 1 cells is gas transfer whilst the Type 2 Pneumocytes produce a protective lubricant, called surfactant.

Figure 3: A diagrammatic representation of an alveolus

Alveoli are the grape-like evaginations of the smallest airways, the bronchioles, encased by a net of very fine blood vessels (capillary net). Via this structure carbon dioxide (CO_2) is transferred from blood into the airway and oxygen (O_2) is absorbed from the air into the blood.



Surfactant

Surfactant (shortened from **Surf*ace* Act*ive* A*g*e**nt**) is a protein rich, fatty fluid that is produced by specialised alveolar cells (Type 2 Pneumocytes) and coats the alveoli like a film. This thin film of fluid protects the alveoli from collapsing and sticking together during exhalation. There are several disorders characterised by the altered production of surfactant and they are collectively known as *Surfactant Metabolic Diseases*. All of these disorders cause a reduction in oxygen uptake, which results in symptoms of heavy and strained breathing. The severity of the disease and symptoms in patients with the same metabolic dysfunction can be variable. Some have mild disease with moderate distress and others very severe disease with severe distress. This group of diseases is a subgroup of **chILD**.

Interstitium

The interstitium (from the Latin for *room between*) is the framework or scaffolding of the lung. It is a made of collagen (a fibrous protein), through which the small blood vessels, nerves and lymph vessels run. Hence it is also essentially the main constituent material of the alveolar walls (***see* Figures 3 and 5**). Therefore, the name *interstitial lung disease* would suggest that this region, the interstitium, is the primary region that is damaged or altered. However, it is important to understand that other structures of the lung (such as the bronchi, bronchioles, alveoli or blood vessels) are often also involved. Furthermore, there are cases, in which not only structures of the lungs are affected, but also of other organs. These cases are termed "*systemic disease with lung* (pulmonary) *involvement*". More common examples of these include rheumatological disorders, which primarily are diseases that affect joints but, not infrequently, can also cause interstitial lung disease.

Typically, interstitial lung diseases result in a thickening of the interstitium. This thickening increases the distance between the alveolar wall and the blood vessels, impairing gas (oxygen and, in severe forms, carbon dioxide) transfer between the airways and the blood. This explains the low oxygen saturations seen in lots of children with interstitial lung disease. Additionally, this interstitial thickening results in the lungs becoming stiffer, making lung expansion and contraction more difficult. Hence, affected children inhale and exhale smaller volumes of air, and then must breath faster to make up for this deficiency. These changes are apparent when the child preforms a lung function test and are described as *a restrictive ventilation disorder*. Within different areas of the lung some parts may trap gas (and be described as *obstructive*) whilst others will have little air in them (*restricted*).

The Blood Circulation

Every organ in the human body requires oxygen to function. The oxygen is collected by red blood cells in the lungs and then transported through blood vessels to deliver oxygen to each organ. The vessels carrying blood from the heart to the organs are called *arteries*, and those returning the blood towards the heart from the organs are called *veins*. The blood returning from the organs is oxygen depleted, and is then pumped by the heart back into the lungs to become oxygen enriched again, before returning back to the organs. This cyclic flow of blood is known as the *Circulatory System*. It is divided into two circuits; *the Systemic Circulation* and *the Pulmonary* (lung) *Circulation* (***see* Figure 4**).

Sometimes the blood pressure in the pulmonary circulation becomes too high (*pulmonary arterial hypertension*, or *PAH*) as a result of a disease altering the lungs architecture (such as interstitial lung disease). This is known as *secondary pulmonary hypertension*, and results in straining of the heart, which significantly worsens the illness. Hence, even if this complication is rare, it is important for patients with **chILD** to undergo regular ultrasound investigations of the heart (echocardiography). This enables early diagnosis and treatment of PAH before it causes significant difficulties.

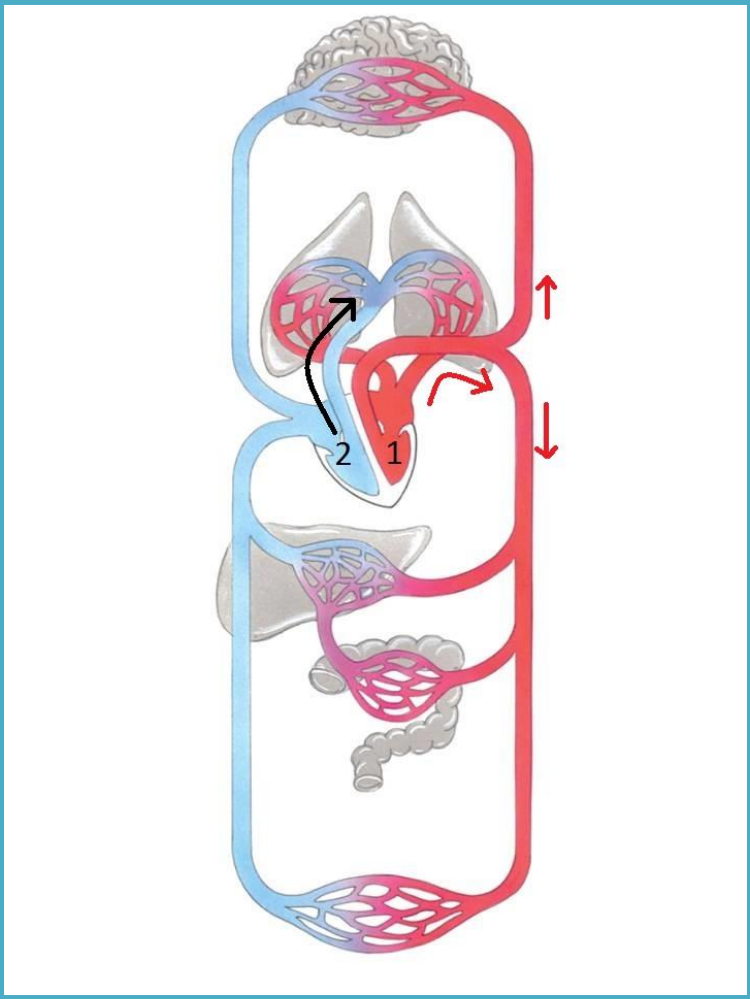


Figure 4: A diagrammatic representation of the Circulatory System

The Vascular Circulation: oxygen rich blood (red) is pumped from the left side of the heart (1) into the body's circulation (red arrows). The oxygen is delivered to the organs and oxygen low blood (blue) returns to the right side of the heart via veins.

The Pulmonary Circulation: oxygen low blood (blue) is pumped from the right side of the heart (2) via the pulmonary arteries (black arrow) into the lung. In the alveoli oxygen is taken up while carbon dioxide is removed. The now oxygen rich blood (red) is returned to the left side of the heart via the pulmonary veins.

CHAPTER 2: chILD

What is an Interstitial Lung Disease? What is chILD?

The term **chILD** stands for **ch**ildren's **i**nterstitial **l**ung **d**isease or interstitial lung disease of childhood. **ChILD** is a collective term for differing diseases, which all share in common pathological changes in the lung's interior support structure, the interstitium (***see* Chapter 1**). Since often not only the interstitium of lung is affected, but also other structures of the lung, doctors have also used the name **diffuse parenchymal lung disease (DPLD).** This alternate naming system is often confusing and misleading, but in the end means the same thing. Imagine a natural sponge (***see* Figure 5**) as being an example of the lung. The much larger empty spaces would be the conducting airways (bronchi and bronchioles) and the very small spaces the alveoli (in which the oxygen is taken into the blood and carbon dioxide is delivered to the airways from the blood). The walls of the small empty spaces would be the interstitium, which are normally ultra-thin and very elastic.

In patients with **chILD**, there is a thickening of these walls, reducing the elasticity and making the transfer of the oxygen and carbon dioxide more difficult. The reduced elasticity means that the lungs (or some areas of the lung) may not expand (*inhale*) or contract (*exhale*) as freely anymore. In terms of a sponge, a normal healthy lung is like a wet sponge, which is easy to squeeze and expands spontaneously to its normal size/shape as opposed to the lungs of a patient with **chILD**, which are more comparable to that of a dry sponge. That is: hard, stiff, difficult to squeeze and unlikely to spontaneously return to their normal shape post compression. The decreased oxygen uptake, results in shortness of breath (*dyspnoea*), and to compensate for this, the patient often breathes faster (*tachypnoea*).

Figure 5:

Using a natural sponge, one can easily demonstrate and understand the structure and function of the normal healthy lung ("a wet sponge"), and then the effects of changes from interstitial lung disease ("a dry sponge").

This increased workload by stiff lungs breathing faster uses lots of energy. Hence these children tire more easily and have a reduced exercise tolerance when compared to their peers (same aged children). Severely sick children need so much energy for breathing, that despite eating sufficiently, they do not gain weight and may even lose weight. The extent of this is naturally related to the severity of the disease.

As **chILD** is in most cases a chronic disease, children may have a reduced lung function in adulthood, but this is not inevitable. But it is not necessarily a one-way-street of worsening function year after year. The contra is often the case. Children can learn to live with their disease, and more often than not (not seldom), can improve year to year with improving exercise tolerance (***see* Figure 6**). Some children even become symptom free over time.

Naturally, the concern for your child's future is enormous and can easily become overwhelming. As such, it is very important to discuss these concerns, since some of them may be easily allayed.

ChILD are rare diseases that tend to affect more boys than girls and occur mainly in children when they are infants or toddlers. Nevertheless, they can and do occur in all ages. Unfortunately, the cause of these diseases frequently cannot be found. For many parents the uncertainty created by this can be very difficult to deal with.

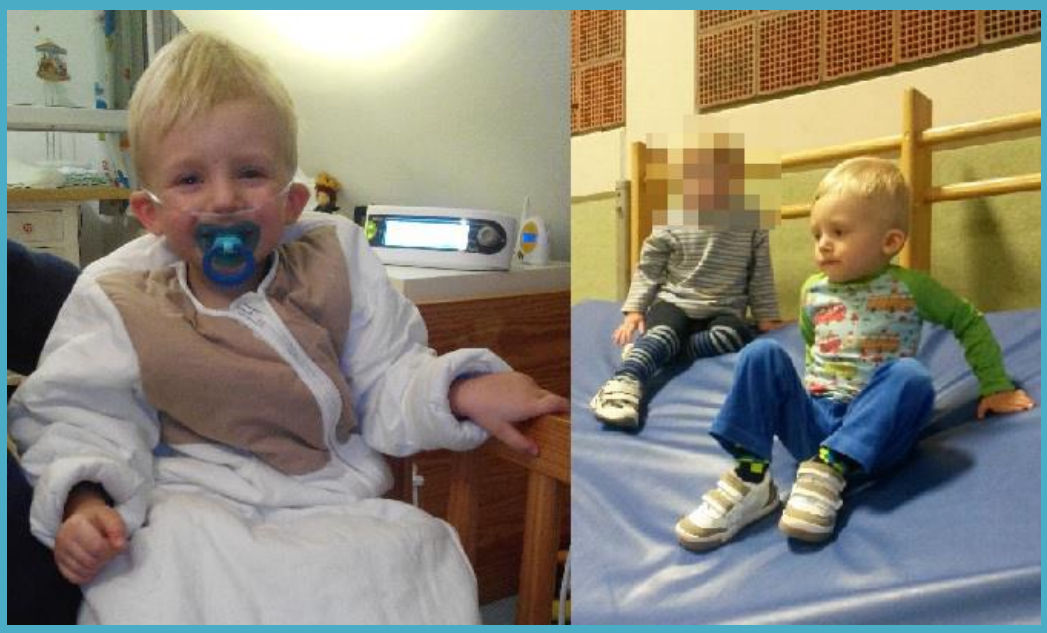


Figure 6:

This is Hanno. He is now 3 years old and was diagnosed at the age of 12 months with ***Neuroendocrine Cell Hyperplasia of Infancy***. He was diagnosed at that time because he had a faster, laboured breathing and was failing to continue to gain weight. The left photo shows him at diagnosis. Through continuous oxygen therapy, he was soon much better. Today Hanno requires oxygen only at night. Otherwise he is doing well and doesn't have problems with his exercise tolerance any more (right photo).

The most precise diagnosis should be attempted by an experienced expert to classify the type of **chILD**. The required investigations must then be performed in a centre with intimate familiarity with these diseases, to obtain the most accurate diagnostic information. In view of the highly variable clinical manifestations of **chILD**, it can also be helpful to have additional expert advisory consultation. Further, the same clinical presentations can have subsequently very different disease progression. So every individual characteristic of your child's disease can be crucial in helping determine the specific type of **chILD**, and hence direct the best treatment strategy for your child.
Finally, one must learn to think in small steps; to take comfort in all improvements, no matter how small, and not to despair with any setbacks.

What are the typical signs and symptoms of chILD?

As previously described, **chILD** leads to more laboured breathing, and in some cases, shortness of breath (*dyspnoea*). The severity of the signs and symptoms in relation to the underlying disease tends to be highly variable. Not every child with **chILD** is necessarily very sick. Some children present with rapid breathing. Other more severely affected children require oxygen therapy (supplementary oxygen). Only rare children are so severely affected that they require artificial ventilation. Not one of their associated signs or symptoms is specific for any one medical condition. That means that most children with fast laboured breathing are not necessarily affected with **chILD**. Since their symptoms are so nonspecific, there is often a long time between initial onset of symptoms and the final diagnosis of **chILD**. It is advisable to present children with the above-mentioned signs and symptoms early on to a specialised medical centre with experience in the evaluation and treatment of children with **chILD**.

Table 1: Common Signs and Symptoms in Children with chILD

- Shortness of breath (***Dyspnoea*)**
- Rapid breathing (***Tachypnoea***)
- Difficult and laboured breathing (***Orthopnoea***)
- Reduced exercise tolerance (e.g. frequent pauses with walking; difficulties climbing stairs)
- Loss of weight, failure to gain weight (***Failure to Thrive***)
- Failure to grow adequately taller, cessation of growth
- Notable breathing noises e.g. rattles, crackles or wheeze
- Persistent cough
- Blue tinged lips (cyanosis) because of low oxygen levels in the blood (***Hypoxemia*)**
- Presence of clubbed fingers and hourglass fingernails (***see* Figure 8**)

Figure 7:

This is Irem, who is now 7 years old. She was diagnosed with ***Congenital Alveolar Proteinosis***. Before the diagnosis was made, she had been very unwell and her parents were concerned for her life. The left picture shows her at the time of diagnosis, when she was two years old. She was very thin, required oxygen therapy and was breathing heavily. With multiple lung washings she progressively improved. The right picture shows her at 4 years of age. She is now doing very well, going to gymnastics club and attending first grade school.

What types of chILD are there?

It is important to know that **chILD** is an umbrella term for a very large group of rare medical conditions, each of which has its own name. All in all, there are approximately 200 different diseases that have been collected under the term of **chILD**. A few years ago, a classification for children was drawn up, to ensure a specific description of the different medical conditions. It was divided into two main groups. One group comprised of conditions that mainly occur in infancy. The second group the conditions occurring in all ages up to adulthood. Both groups are then further subdivided into further subgroups. The conditions are listed together in each subgroup, based upon similar development. It would go beyond the boundaries of this booklet to attempt to list and document all 200 subtypes of **chILD**. You will need to discuss with your treating doctor the specifics of your child's type of **chILD**. Nonetheless, there are many similarities amongst these different types; hence this handbook should be able to provide answer for some of your questions.

Figure 8: Finger clubbing and hourglass nails

Both photos are showing a boy with clubbed fingers and hourglass nails. They are suggestive of an underlying chronic oxygen deficiency and can develop in children with severe lung or heart conditions. The striking spoon-shape of the fingertips is called "clubbing" (a), the broadening and rounding of the nails is called hourglass-shape (b). Often these changes will coexist. This boy was diagnosed with ***pulmonary fibrosis***. The cause of this remains unknown. For years he was mistakenly thought to have bronchial asthma. These types of changes are never present in asthma. After diagnosis with pulmonary fibrosis, he was commenced on pulsed cortisone therapy. Additionally he was given hydroxychloroquine and azithromycin (***see* Chapter 4**). Today the boy is much better, with regression of both the finger clubbing and hourglass nails.

What are the severity grades of chILD?

The severity of the illness can vary significantly, even in children with the same diagnosis. Some seem barely affected beyond having a slightly faster breathing rate. Many children require temporary oxygen therapy during some stage of their disease progress. For cases with severe breathing difficulties, children may require more active assistance with artificial ventilation (***see* Figure 10**). In the very rare and the most severe cases, children may die of their condition or only survive by having lung transplantation. The American paediatric lung specialist, Dr. Leland Fan, has proposed the following severity classification system for children with **chILD**:

Mildest Form

No Symptoms (asymptomatic)

Symptoms but normal blood oxygen saturation levels

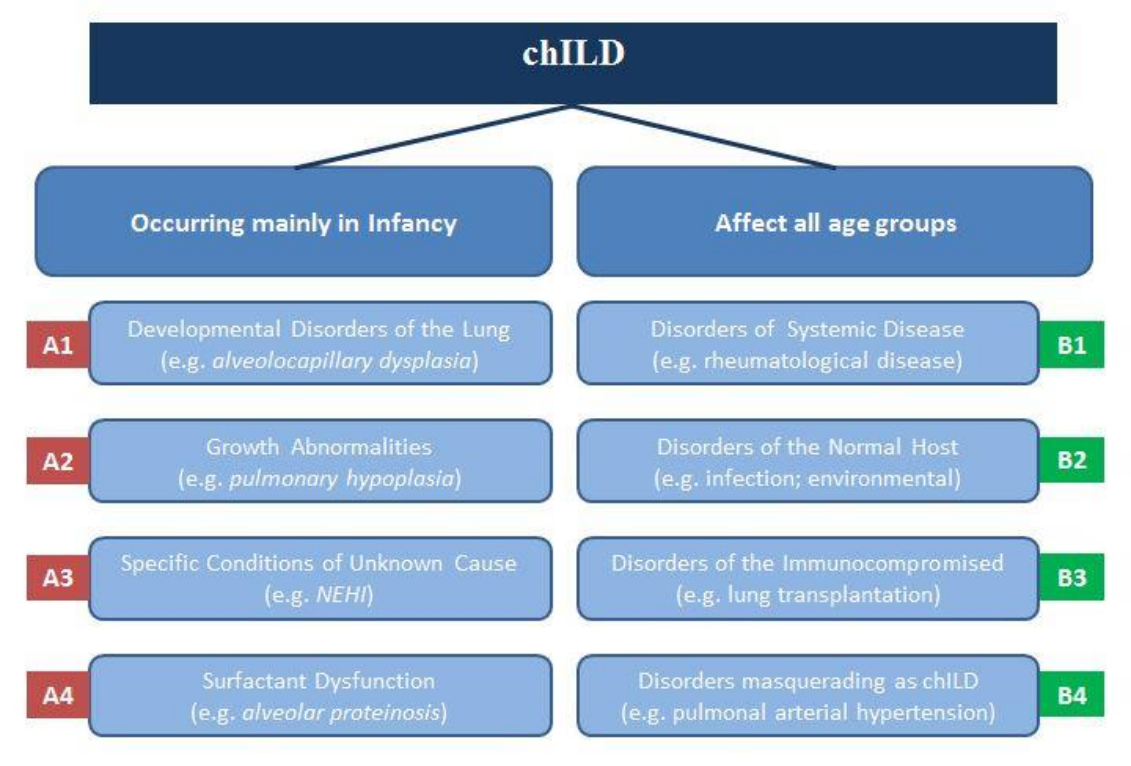
Symptoms with decreased blood oxygen levels

Symptoms with decreased blood oxygen levels at rest

Presence of Pulmonary Arterial Hypertension (***see*** **Chapter 1**)

Severest Form

Figure 9: Classification of chILD



Can chILD be inherited?

Yes, there are inheritable forms of **chILD.** The presence of recurrent severe or chronic lung problems in relatives outside of the immediate family can be suggestive of this. When the parents are closely related (consanguineous), the risk of inherited disorders drastically

increases. These days it is possible to screen for some of these disorders with genetic blood tests (***see* Chapter 3**).

Figure 10:

This is Enes, who is now 3 years old. Since birth he was oxygen-dependent. At 1½ years old he had to be ventilated because of severe ***alveolocapillary dysplasia***. As seen in the left photo, he nonetheless insisted on learning to ride a tricycle. Visible behind him is the tubing for the ventilator that he required at the time. Subsequently Enes received a new (transplant) lung and is now doing as well as other children of his age (right photo). He no longer requires either ventilation or oxygen therapy.

CHAPTER 3: DIAGNOSIS

How is the diagnosis made?

When the suspicion of interstitial lung disease is first raised, optimally your child should be seen at a specialist centre with expertise in the evaluation and management of these rare disorders. Now of course the question arises, where to find such a specialised centre. Visiting the chILD-EU website can assist with this endeavour (it is written in English). There are email links provided for obtaining patient information in the countries of United Kingdom, USA, Germany, France, Turkey, Italy and Spain. The link is: **http://www.klinikum.uni-muenchen.de/Child-EU/en/care/patient_information_service/index.html**

How can I optimally prepare for the appointment in the centre?

An essential component in the successful diagnosis of your child is through the taking of a detailed history. It will not only be asked: what symptoms are present, when the symptoms started, what investigations have been performed, and what treatments have been given so far; but also if and how often has your child been treated in hospital. Therefore, it is beneficial to prepare, as chronologically correct as possible, a list (diary) with this prior medical history and bring it with you to the first conversation at the centre. Attempt to ensure that copies of all previous medical letters and results of previous lung function tests accompany you and your child to this first visit. As well, you should ask for all prior radiological imaging (x-rays and CT-scans) to be burned onto a CD and bring these as well or alternatively transferred electronically to the clinic. Some of the signs or symptoms may manifest only intermittently (and never with a doctor present) and may be difficult to subsequently describe. In this setting, having previously filmed your child when they are manifest and bringing these recordings to the visit would be invaluable.

Could it be something other than chILD?

Yes, since the symptoms and signs in children with chILD are non-specific. That means that there are many other conditions that could be the potential cause of your child's medical illness. Therefore, an important part of a diagnostic workup is to exclude these other conditions.

ATTENTION

If you already have an appointment in a specialist centre, or it is foreseeable, ensure that all the special investigations (e.g. CT-scans of the lung; investigations under anaesthesia especially bronchoscopy and lung biopsy) required are performed only in that centre. This is because investigation results from a centre experienced in chILD will be more meticulous and reliable following the investigation guidelines on the ChILD EU website. Hence, as these investigations are often very stressful for your child, it would be undesirable to repeat them because of indeterminate results.

Table 2: Some conditions with a Similar Presentation to chILD

- Congenital Malformation of the Airways
- Congenital Malformation of the Lung
- Congenital Heart Disease
- Bronchial Asthma
- Chronic Infections of the Airways (including choking on food and aspirating)
- Immunodeficiences
- Cystic Fibrosis
- Functional Disorders of the Cilia (e.g. ***primary ciliary dyskinesia***)
- Idiopathic Pulmonary Hypertension

Which investigations are required?

In the following section we will describe and explain the most commonly utilised investigations in the workup of your child's diagnosis. Certainly, there are other investigations or procedures utilised. Their lack of presence here does not equate to them being unwarranted, rather only that they are more rarely performed with the majority of cases. It is your right to ask the doctor why a certain investigation shall be done, what they are hoping to discover and what potential risks there are with it. Finally, after each investigation, do not hesitate to ask your child's doctor(s) to show and explain the results of all these investigations.

Basic Investigations

The investigations listed in **Table 3** should principally be done in all children suspected of having a **chILD.** They are therefore called the basic investigations. Their main advantages are that they tend to have little impact on your child and deliver rapid results (which help determine subsequent investigations).

Table 3: The Basic Workup for Children suspected of having chILD

- Detailed History Taking
- Measurement of Body Weight and Height
- Thorough Clinical Examination
- Measurement of Respiratory (*breathing*) Rate
- Measurement of the Oxygen Saturations
- Blood Gas Analysis (usually capillary sampling for carbon dioxide analysis)
- Plain Chest X-ray
- Lung Function Tests (usually only in children over 5 years of age)
- 6 Minute Walk Test (if only over the age of 5)
- Echocardiogram (ultrasound of the heart)
- Sweat Test (testing for Cystic Fibrosis)

Plain Chest X-Ray

A simple X-ray can provide the doctor with rough information regarding the organs within the chest (thorax), including heart, lung and the great blood vessels. Some changes in the X-ray can support the suspicion of an interstitial lung disease, but will not prove or disprove it. The advantage of this test is that it is fast, pain free, gives only a very small dose of radiation (***see* Table 4**) and does not require sedation. The images are available immediately and can be reported by the doctor. Its main disadvantage is that it cannot demonstrate subtle or fine changes. One can image that it is like looking through a milk glass window at a garden; outlines of trees and plants are clearly seen but more detailed features of the plants (like the pattern of the leaves) are not visible.

Figure 11: Comparison of an X-ray and a CT-scan

These two photos help demonstrate the difference in the level of detail given by a plain X-ray and a CT-scan. The photo on the left mimics a plain X-ray and you can guess that you are looking at a leaf with a lighter structure in the middle. That this structure is indeed a water droplet is only apparent with the right picture (mimicking a CT scan).

Computer Tomography of the Lung

In contrast to plain X-rays, CT-scanning or computer tomography shows a detailed image of and fine alterations in the lung, heart and blood vessels. To continue with the milky glass analogy, the CT scan can be compared to looking through a clear glass window, where all the fine details are easily discerned (***see* Figure 11**).

The CT-scan is the most important radiological imaging technique for suspected **chILD**. Although it is unusual for it to establish the precise diagnosis, the images confirm the presence of ChILD and help plan the best site to obtain tissue samples. A disadvantage is the higher radiation dose (***see* Table 4**). Therefore, repeated imaging because of poor image quality should be avoided. Clearly, one significant factor in determining the image quality is the quality of the scanner; another is that of movement artefact (i.e. movement related blurring of the image). Although a CT-scan is pain free and only takes seconds, any movement (including breathing) can result in the images becoming hazy and making fine details unclear. So before taking the images, your child will be asked to take a deep breath in and then hold it, to see if they are able to. Furthermore, making this experience more fun and familiar for your child will enhance the likelihood of success. One example is utilising playfully practice with your child the day prior to the investigation; by making a game of them trying to lie still, hold their breath in, and then exhale and hold the breath out. Additional benefit comes from visiting the procedure room the day prior to the scan, and explaining the sequences involved with the test in an effort to make it less intimidating for them.

Infants and young children often require a general anaesthetic to obtain the good quality images required to help diagnosis. This is also the case for very sick children who are often not able to either hold their breath or to lie still long enough during the scan. Centres sometimes aim to perform other investigations that also require anaesthesia at the same time (e.g. bronchoscopies). In some cases, a contrast medium (radio-opaque dye) may be required to be injected into your child. This can cause allergic reactions or problems with thyroid dysfunction in rare cases. Should your child have a history of either of these, it is important to let the doctor know. Finally, as with all investigations, make sure that you know why the test is needed, trying to demonstrate and its potential complications/risks. The better you understand what the test involves and why it is required, the easier it is to consent to and support your child to undergo it.

Table 4: Radiation Exposure comparisons with Radiological Imaging

<u>Comparative Radiation Dose:</u>	
Natural, yearly background radiation exposure:	2.7 – 6.2 mSv/year
Transatlantic Flight (8 hours):	0.04 - 0.1 mSv
Plain Chest X-ray:	0.01 mSv
Thoracic (chest) CT-scan:	2.0 - 2.5 mSV

Lung Function Testing

Lung function testing (***see* Figure 12**) is a non-invasive test that gives valuable insight into the type and functional impact of lung disease. As it is technically demanding, in the sense that the child needs to be able to understand and follow sequential directions, for many but not all tests it is usually only performed in children from the age of 5. Infant tests are offered in very centres and there is much less experience in interpreting these in ChILD. The unique information it can provide means that it makes sense to practise lung function testing in the outpatient setting from the age of 3. If children need to practice they can take home a peak flow meter – though the peak flow measurement itself isn't of value in ChILD (***see* Fig 13**).

Figure 12: Lung Function Test

Lung Function Test method: a nose clip is applied so that the child only breathes through their mouth. The child fully encloses the mouthpiece with their lips, without biting onto it. Typical directions given during the test are: *"Please breathe slowly in and out. Now breathe out slowly and fully until there is no air left in your lungs. And now take a breath in as deeply as you can and then exhale out as fast as*

Figure 13: Peak-Flow-Meter

This is a common Peak Flow Meter where lung function testing can be practiced, as well as being a basic monitoring function at home. Although these devices are imprecise, through regular measurements, it is possible to establish an individual's average "healthy" peak flow value. Daily monitoring can help demonstrate when there are deteriorations in function, as can happen

Blood Gas Analysis (BGA)

This test is primarily used to measure the partial pressures of oxygen and carbon dioxide in the blood. Since **chILD** can reduce both the uptake of oxygen into and the elimination of carbon dioxide from blood, this test can help assess the severity of the lung disease. Oxygen levels require an arterial blood gas – these are painful and not often performed. Measurement of carbon dioxide is the most helpful part of this test and usually this is done by taking a capillary sample of blood, usually from the tip of a finger.

Measurement of Oxygen Saturation

A non-invasive, non-painful but slightly less precise way of measuring arterial blood oxygen levels is done with a superficial skin probe *(pulse oximeter)*. Oxygen saturation monitoring is easy to use and so provides most information for oxygen levels in ChILD. It can be attached to either a finger or ear lobe (with a plaster) and then connected to a monitor via a cable. This can show both the oxygen saturations and also the heart rate (***see* Figure 14**). Some monitors can store these measurements over a period time (e.g. 24 for 48 hours), for later review and analysis. Children who require night-time or continuous oxygen may have a pulse oximeter at home, but this isn't always necessary.

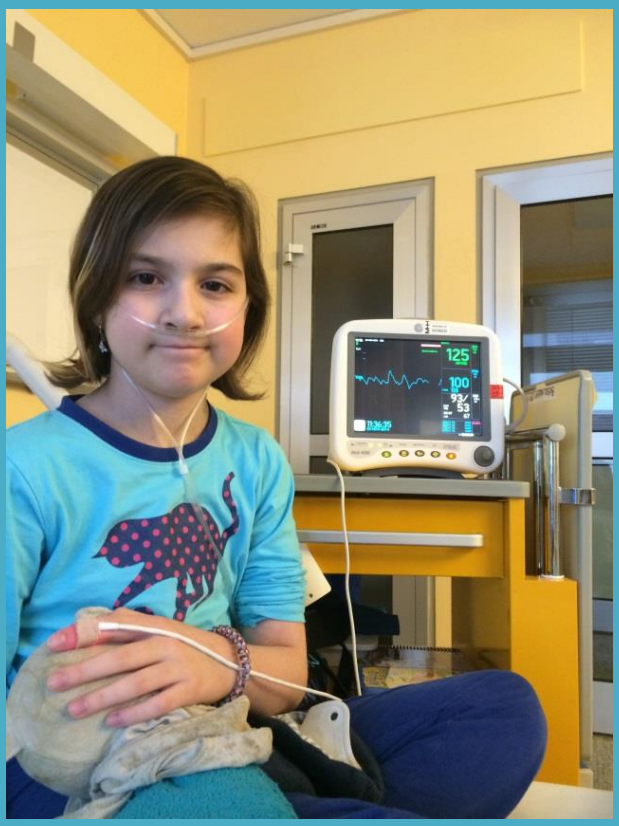


Figure 14:

This is 11-year-old Cara. After having radiation therapy for a rib tumour, she developed pulmonary fibrosis and requires oxygen therapy. The pulse oximeter probe is attached to her left index finger with a plaster. The monitor behind her is showing her heart rate (green) and oxygen saturations (blue).

Genetic Testing

Only in more recent times can a few types of **chILD** be diagnosed directly with genetic blood tests. These tests can take days to weeks to come back. Nonetheless a definite advantage of them is that they give a clear, precise diagnosis without the need for lung biopsy (when positive). Unfortunately, though only a few types of **chILD** can be diagnosed this way:

- Surfactant-Protein-B Deficiency (SPB-Mutations)
- Surfactant-Protein-C Deficiency (SPC-Mutations)
- ABCA3-Deficiency (ABCA3-Mutations)
- Alveolar Capillary Dysplasia (FoxF1-Mutations)
- Brain Lung Thyroid Syndrome (TTF1-Mutations)
- Congenital Pulmonary Alveolar Proteinosis (CSFR2A, CSFR2B-Mutations)

Further Blood Tests

During the initial evaluation, your child will have a large number of blood tests done and many of them will be repeated. Beyond genetic tests, blood tests don't usually confirm or exclude a diagnosis of **chILD**. So you would then be right in asking why we perform so many tests. The answer is because the results can offer important indicators as to which disease it may or may not be (including the exclusion of some non-chILD causes of the symptoms). Each single result is like a small piece of a large, complex jigsaw puzzle (one of 200+ different jigsaws). This, as you could imagine helps explain why establishing the diagnosis can be so difficult.

Bronchoscopy

Bronchoscopy is a procedure in which the investigators are able to look directly inside the lung. This is done by introducing a thin cable (2.8-5.8mm outer diameter), with a light source at its tip, directly down the airways and watching the images through it on a screen during the procedure (***see* Figures 2** and **15**). It is possible to visualise much of the upper airways (pharynx, epiglottis and larynx) and the lower airways (trachea and bronchi) during the procedure. During the passage of the cable, the airways are examined for any malformation, being too floppy/soft or narrow, excessive secretions or signs of inflammation (irritation of the surface of the airways). Normal saline solution (*salty water*) is flushed into a lung lobe, sucked out and then sent for further testing. This is called a *Bronchoalveolar lavage* (BAL). Samples from this are tested for bacteria, viruses and inflammatory cells. Additionally, in special cases, washed out surfactant will be evaluated for abnormalities. Infrequently, small special forceps can be introduced via a working tube down the cable to obtain small tissue samples (*biopsies*).

Bronchoscopy is always done under anaesthesia in children and young persons. As such it does not cause pain or distress for a child. During the procedure some children may only require supplementary oxygen, breathing independently, whereas others may need to be ventilated (via special masks or breathing tubes). Furthermore, the children are continuously monitored during the procedure for any deterioration. Severe complications are very rare. Commonly, children may have a mild temperature, cough or be hoarse on the day of the procedure. Seldom, they may require additional supplementary oxygen with intensive care

monitoring. Finally, minor, non-dangerous coughing up of blood (*haemoptysis*) occurs sometimes post procedure when biopsies were performed.

Figure 15: Photo of a Bronchoscopy

The doctor doing the procedure (right side) is holding the bronchoscope in her right hand. On the monitor (left side) she is evaluating the airways (here the trachea) while passing down the tube.

Lung Biopsy

The lung biopsy is the most important investigation in the diagnostic workup of suspected **chILD**. It becomes mandatory after other possible conditions have been excluded and when the previous investigations have failed to provide a definitive diagnosis.

Today most biopsies are done via a minimally invasive procedure known as *VATS*, or *video assisted thoracoscopic surgery* (***see* Figures 16** and **17**). In this procedure instruments are introduced into the chest cavity, under anaesthesia, via three small incisions. The surgeon can look directly at the lung via these instruments and then take small biopsy samples (***see* Figure 17**). These samples are usually around 0.5-1cm by 0.5-1cm big and their loss has no impact on your child's lung.

The alternative is an *open lung biopsy* where a larger skin incision is made (3-5cm), through which the samples are taken. Advantages of the VATS procedure over the open biopsy include: less post-procedural pain; chest drains are required less often (sometimes required for secretions or air extraction); earlier discharge home; and subsequently smaller (barely visible) scars.

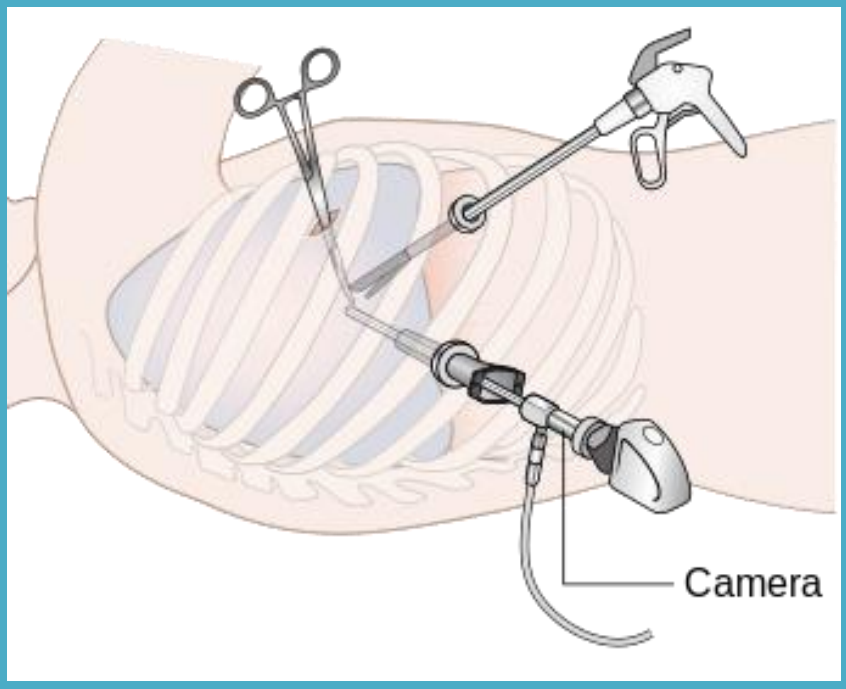


Figure 16: Diagram of a video assisted thoracoscopic surgical (**VATS**) lung biopsy

Through the inserted camera (bottom) the surgeon can obtain a detailed look of the lung's surface. Then a biopsy can be done with two additional instruments introduced through two more small skin incisions. The child is under anaesthesia and does not feel any pain during the surgery.

The Diagnosis of chILD has been made. Now what?

Despite having acknowledged the possibility that your child may have a chronic lung disease during the initial conversations, there has always remained the hope that the investigations will demonstrate no major problems and that all is fine. Thus when the diagnosis is confirmed, it is still often difficult to deal with. Thoughts race through your head and sorrow for your child's misfortune constricts your heart.

In the following chapters we will focus on how to deal with this. We will approach this by utilizing frequently asked questions from other families with affected children, trying to find answers and approaches for solutions. Ultimately though, every individual and every family is unique. This often requires differing solutions that can only be achieved by discussing your needs and problems with your treating team. It is extremely important to share your anxieties,

sorrows and problems. Allow others to help you. It is not a sign of weakness or failure, on the contrary!

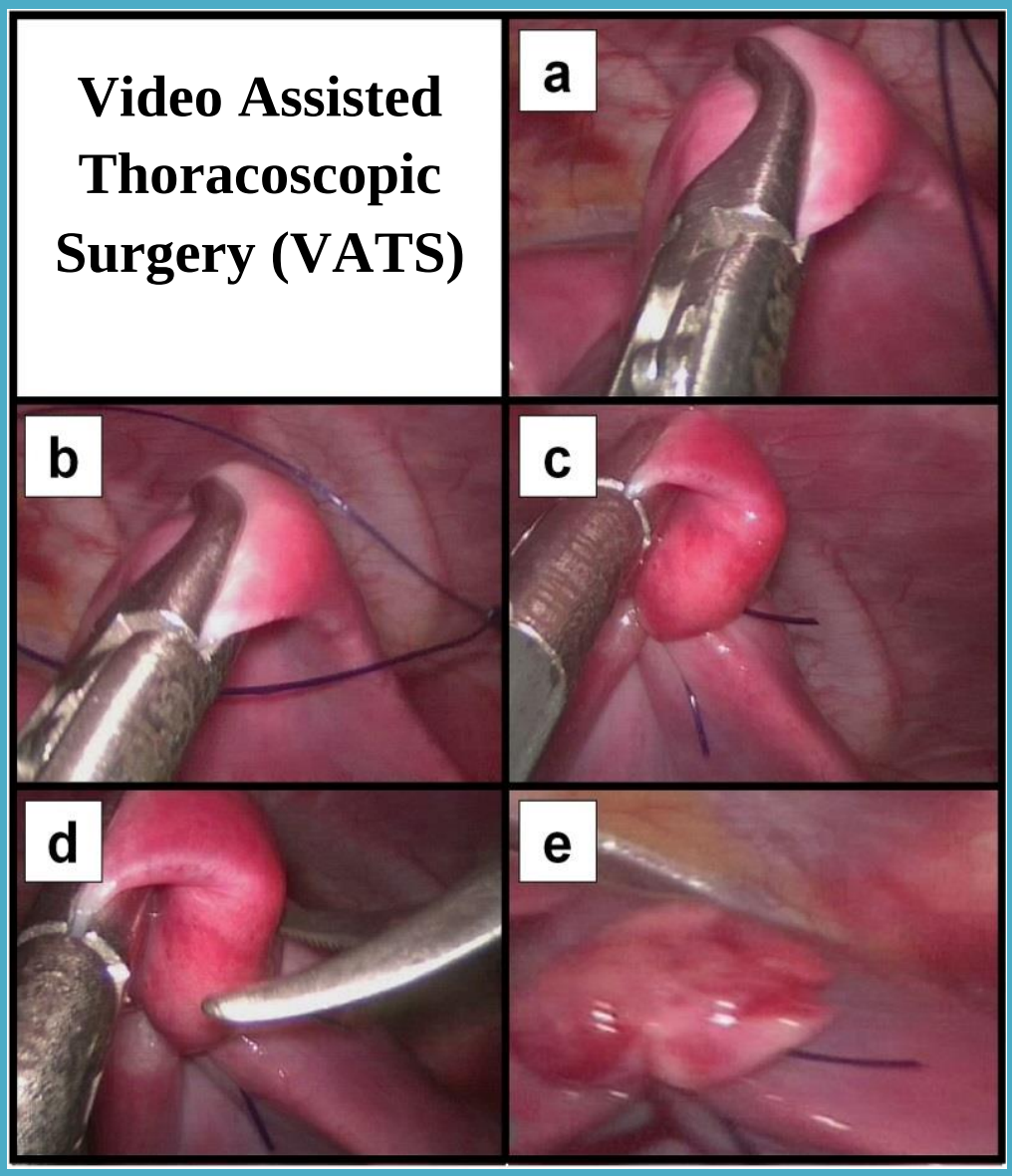


Figure 17: Photos of a **Thoracoscopic Lung Biopsy** procedure (reproduced with the kind permission of Prof Dr med. Ure and PD Dr med. Dingemann, Medical University Hannover)

a) Selection and clipping of suitable lung area
b) Positioning of a noose (snare) around the sample to take
c) Post tightening and knotting the noose
d) Excising the sample from the lung with scissors
e) Sample removed. The tightened noose acts as a tourniquet stopping bleeding and air leakage from the lung

The Diagnosis Conversation

Allow yourself to hear exactly what medical condition your child has. Request clarification of anything you have not understood. Share what your fears are with this diagnosis. In most cases the fear of your child dying can be alleviated. Unfortunately, the question of how your child will be in a week, a year or ten years cannot usually be answered. Often people forget questions that they want to ask their doctor during an appointment. Thus, it can be very helpful to write down a "shopping list" of questions before you speak with them.

Figure 18:

This is Soraya, who is now 8 years old. Until her 7th year of life she was completely healthy. After an infection she developed worsening dyspnoea and deteriorated daily. A lung biopsy established a diagnosis of **chILD**. The actual cause remains unknown, which has proven very difficult for her parents. The left photo shows Soraya shortly after the biopsy. She was unwell, on supplementary oxygen and required a chest drain for excessive wound secretions and leaked air in the chest cavity. Subsequently she underwent treatment with cortisone and azithromycin, with which she happily improved. The right picture shows her several months post the biopsy. At which point she no longer required supplementary oxygen, was going to school and roller-blading (despite having incomplete recovery in her lung functions).

Should I have recognised things earlier?

This question can be answered with a resounding "No". The symptoms often develop insidiously. These slow, progressive signs and symptoms are incredibly difficulty to notice from day to day (which is especially true for most parents who see their child every day), and are far more apparent when you see the child only sporadically. Furthermore, the signs and symptoms are very non-specific. What parent has not had their child at some stage tired, moody or lacking energy? Moreover, these diseases are incredibly rare, with very few doctors

having ever encountered them and many have not even heard of them. There are many more common and less concerning (often harmless) causes/reasons for these complaints. It is nigh impossible to reproach your general doctor/paediatrician in this setting, let alone yourself.

Have I done something wrong?

Again this question can be answered with a definite "No". On the contrary, you have done everything right. The only reason this rare condition has been successfully diagnosed in your child, is because you and your doctor recognised and acted on the signs and/or symptoms. No type of **chILD** has been caused as a result of the wrongdoings of the parents.

How do I explain this condition to my child?

We will work together to explain what this condition is to your child, in an appropriate way for their age and at a pace consistent with their level of understanding. It is sometimes helpful to use uncomplicated pictures and diagrams with the explanation, particularly in younger children. Not infrequently, children will be very closed or withdrawn during these conversations. This does not reflect that they do not understand what is being said. Commonly, children will ask their questions after the doctor leaves the room, and it is important to write them down, so as to address them at the next visit with the doctor. Sometimes, children cannot clearly put into words things that are weighing them down. Here, art therapy can be helpful. It is amazing how unbelievably clearly children can paint their thoughts, anxiety and sorrows. Art therapy also helps children confront their disease and lose anxiety about it. Children have a right to know their diagnosis and it is important not to try to protect the child from it. They will know better than anyone else that something is wrong with them and that they are unwell.

How do I explain this to my family now?

It can be very useful to have all the family members involved with the care of the child, present together at the diagnosis conversation. Otherwise, it may be possible to arrange an additional appointment with the doctor, to explain things a second time. Or in the case that someone cannot attend at all, it can be helpful to bring a list of their questions to the doctor and jot down the answers to inform them later.

How do we need to restructure our daily life?

This very much depends on the individual family and the severity of your child's illness. There are a number of services available, both financial and social (such as household aid, medical prescriptions, transport, and homecare) which can reduce the strain on your family. These are dependent upon many things including: where you live, severity of disease and family structure. Common questions include: "Who do I ask?"; "What am I entitled to?" and "What forms do I need to fill in?". Your greatest resource in resolving these issues is often a Social Worker from one of the larger hospitals. There are more detailed notes regarding this subject and the available resources in **Chapter 10** (with the most important contact addresses).

How is this reconcilable with work?

Today this is an even more important question, as often both parents are working. Again the answer here is dependent on the individual family's situation, nature of the parents' employment and the severity of the illness. It should also be mentioned here that most children with **chILD** lead relatively normal lives despite their disease. Thus, **chILD** does not mean one or both parents must stop work. Nonetheless, many cases require episodes of prolonged hospitalisation with increased parental support of the child. During these episodes it advisable to discuss things with your employer, as often they will be very supportive and helpful in these situations. Advice from a Social Worker could again be invaluable here.

Will my child become stigmatized or socially isolated?

This is a legitimate and frequently raised concern. Your child will not have limitless energy like they did before the disease. Most can still play around, go on class trips and play club sport, but likely not to their pre-sickness level. On top of this exercise limitation, your child may have to deal with teasing from children of the same age because they are weaker or are treated differently by teachers.

It is essential to give your child the chance to continue living a relatively normal life, with as few limitations as possible. The diagnosis of **chILD** does not equate to an end to sport or exercise for the majority, only in the extreme cases is it potentially so. Sport is not dangerous for children with **chILD**; rather it can be an important component of their treatment. Children are excellent at self-regulating themselves with exercise, in regards to stopping when they have reached their limit. Thus, you do not have to be worried about them over straining themselves and exacerbating their disease process. Likewise, it is important that adults do not push these children past their limits or into doing activities that they do not wish to. Talk to your doctor if your child is not able to do sport in a normal sports club because they have severely reduced exercise tolerance. There are many alternatives, including playing sport in one of the many groups for children with disabilities or joining a non-sporting club (e.g. music, handy crafts etc.). There are the benefits beyond just fitness, including developing self-esteem through the promotion of individual skill and teamwork.

Teasing by other children is rarely through malice, but tends to be more through ignorance. In addition, there can be evident uncertainty or insecurity from some teachers or carers when they deal with children with chronic conditions, which the other children will intuitively pick up on. Occasionally, the source of being rejected is from an irrational fear that the medical condition could be/is contagious. All of these can be eliminated through good education, and some specialist centres are able to offer doctors or nurses of other healthcare professionals to attend the school or nursery to carry out this educational work.

CHAPTER 4: TREATMENTS

Who will be treating my child?

The most important member of the treating team for your child is you, their parent. You know your child the best and spend the most time with them. You were the first to recognize that they were unwell and are coordinating all of their medical appointments. But you are not alone. Your team is not just the specialist (expert) doctor, but also multiple other professionals (***see* Table 5)**. Not every treating team will be the same, or stay the same over time. Each individual member of your team will have input depending on the requirements of the specific disease type, time course of the disease and the individual needs of the child and their parents.

Table 5: The Treating Team

- Your **local paediatrician** will remain your primary carer and continue to be responsible for the basic medical needs of your child.
- The **paediatric pulmonologist** (specialist doctor for children's lung diseases) is responsible for establishing the diagnosis and treatment plan for your child. They will coordinate the involvement of all the other professionals required to manage the disease. There should be no change in treatment without their approval.
- The **paediatric cardiologist** (heart specialist for children) will perform echocardiograms at regular intervals to exclude the presence of pulmonary artery hypertension (***see* Chapter 1**).
- The **psychologist** is a very important member in the team, supporting the child and family in dealing with (and overcoming) the stresses of the underlying condition. They develop a special trust relationship with the family, dealing with the non-medical aspects of the disease in far more detail than the doctors.
- The **paediatric surgeon** will become involved if a diagnostic lung biopsy is being considered.
- Situationally required specialists include **rheumatologists, immunologists, intensive care physicians, gastroenterologists** and **human geneticists**.
- The **dietician** provides advice regarding optimal nutrition. Assessing the caloric intake and correcting as required.
- **Physiotherapists** are very important in the treatment of **chILD**. They teach special breathing techniques to help mobilise lower airway secretions if present (autogenic drainage), help improve physical conditioning and fitness (i.e. exercise tolerance).
- **Logopedists or Speech Therapists** support and assist children that require tracheal cannula with alternate speaking techniques.

Table 5: The Treating Team (Continued)

- **Social Workers** assist with contacting and dealing with various offices including health insurance and dealing with their required paper work (e.g. applications for financial assistance, rehabilitation or special medical equipment).
- **Nurses** play an important role in the treatment of the children as inpatients. They have the closest contact to them and are therefore, in addition to their medical care, an important person of reference.
- **Portage or play therapists** not only help children to express themselves through creativity, but also some hospitals may also have access to **sports therapists, hospital clowns** and **hospital teachers.** These team members help to improve physical health and wellbeing, and also help them cope better with their disease, education and break up the tedium of their hospital stay.

When will my child need oxygen?

ChILD reduces the uptake of oxygen in the lung's alveoli (***see* Chapter 1**), the extent of which is relative to the severity of the disease. The body is dependent on having sufficient levels of oxygen in the blood to maintain function. When these levels become too low (*hypoxemia*), the person may develop the symptom of feeling short of breath (*dyspnoea*). Some children compensate for this lower oxygen uptake by breathing faster and deeper. For others with more severe disease, this mechanism is inadequate and no longer compensates for the reduced uptake. This is known as *respiratory failure* or *insufficiency*. Supplementary oxygen in the inhaled air is required to correct the deficiency in these cases. Normally, air at sea level consists of 21% oxygen, which can be increased, depending on the type of delivery, up to 100% with supplementation. Some children may require oxygen therapy continuously, whilst others only intermittently (e.g. while asleep, during physical exercise or infections). The amount required is determined as an inpatient, by titrating the concentration (percentage of oxygen) up or down while usually by measuring oxygen saturations (pulse oximeter, ***see* Chapter 3**)

Chronic oxygen deficiency can result in the child having a limited exercise tolerance, feeling tired and lethargic, not gaining weight or even losing weight. In short, they are unwell. More concerning is that this chronic hypoxemia may lead to *pulmonary arterial hypertension* (high lung circulatory pressure, ***see* Chapter 1**), which puts strain on the heart. Most of these signs, symptoms and complications can easily be treated with supplementary oxygen. Thus oxygen is the primary and optimum treatment for all children with respiratory insufficiency.

My child requires oxygen: Now what?

Essentially you will not need to do anything; thankfully it is someone else's responsibility. Your child's healthcare professional will provide you with the prescription for the oxygen and all the required equipment. Then they may refer you to an oxygen specialist nurse who will liaise between yourself and one of the many oxygen supplier companies. Subsequently,

arrangements can be made to arrange delivery of the equipment to your home. Naturally you will receive a detailed education about the safe use of this equipment. This education will sometimes occur while your child is an inpatient, especially if your child requires the oxygen to be present at home on their day of discharge.

It is necessary for everyone on oxygen therapy to have his or her saturations monitored (continually or periodically) to ensure satisfactory oxygen saturation. Part of the supplied equipment may include a pulse oximeter (***see* Chapter 3** and **Figure 14**). Please discuss with your child's clinician whether you require a monitor at home. When the oxygen requirements have been the same for a while (*plateaued*) and your child is relatively well, there is no need for continuous daily monitoring. Your doctor will decide whether or not you will need to monitor your child's oxygen saturations during sleep. There would be little value in monitoring your child's saturations without the monitor being able to let you know when the saturations are too low. As such each device has an inbuilt alarm, which can be programmed to activate whenever the saturations fall beneath a predefined (*baseline*) level. Normal saturations can transiently fluctuate up and down, and any transient dips beneath this baseline level will set off the alarm. These transient dips can be seen as false alarms in the setting of a potentially too tightly set baseline level. This can be nerve wrecking for anyone, especially light sleepers. Luckily this level can easily be adjusted, although no changes should be made without the approval of your treating doctor.

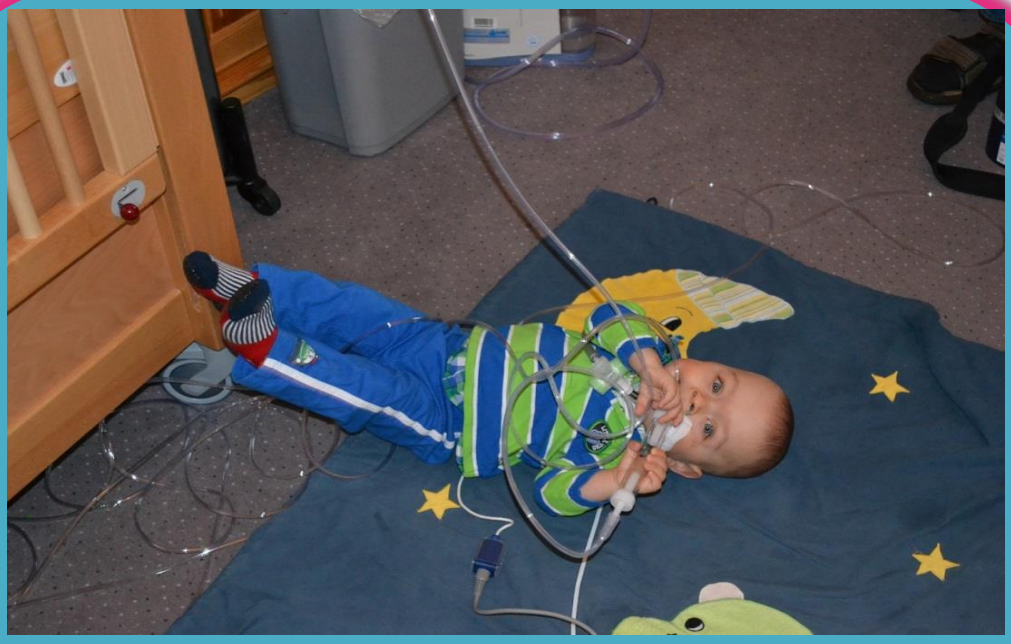


Figure 20:

This is Joris (also ***see*** **Figure 24**), an extremely lively and active boy despite requiring continuous oxygen supplementation. The addition of the "long line" meant he was able to move around relatively freely (under close supervision) at home.

Oxygen Concentrators

Oxygen concentrators are the most commonly used equipment that can be used for long term oxygen therapy. They can deliver close to pure (100%) oxygen to your child by sucking in environmental air and filtering out all the other gases. They are powered by electricity and thus always require an electric supply to function. There are portable battery operated systems (e.g. Inogen One®, Sequal Eclipse®) that can last for up to 8 hours. These oxygen systems are relatively light, as they filter the oxygen out of the air so they don't require heavy oxygen cylinders. Nonetheless, with oxygen dependent patients, it is always necessary to have an emergency oxygen tank at hand in case of electricity failure. Another weakness is that the flow rate with currently available models is limited to 8 litres per minute, although it is only rarely that children require a rate greater than this.

Liquid Oxygen Systems

When oxygen is cooled to -183^{o}C it changes from a gaseous state into a liquid. One litre of liquid oxygen is equivalent to 850 litres of gaseous oxygen. Hence, a single oxygen tanks can last a significant amount of time and further they do not dependent on electricity to function. Small transportable liquid oxygen systems can repeatedly be refilled from a larger and heavy storage tank at home. This gives the child greater mobility and freedom to move about. These

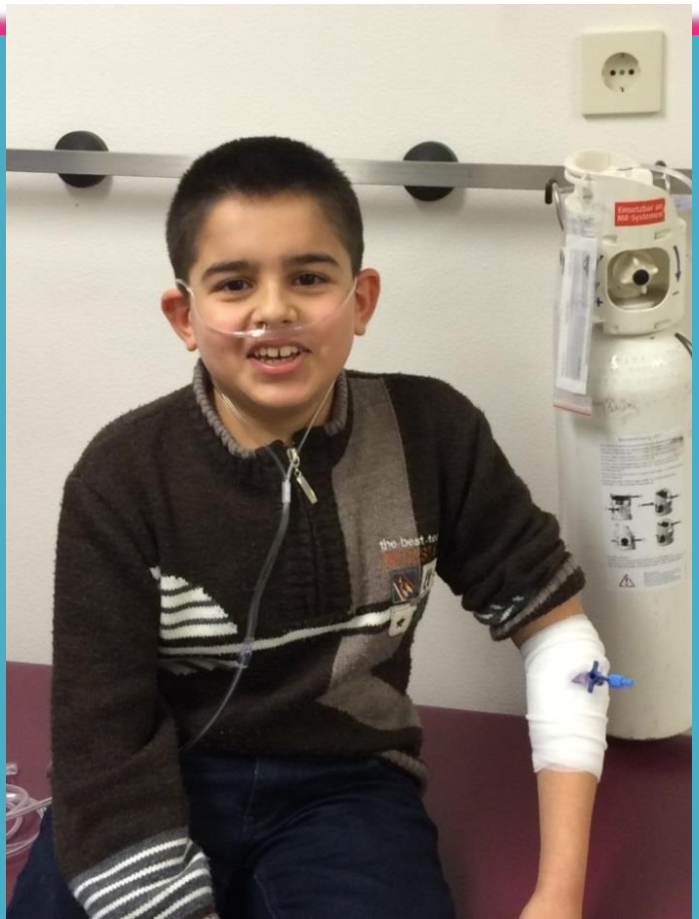
systems are noticeably lighter than and have a greater storage capability when compared to gaseous tanks of the same size. However, it should be noted that liquid oxygen should be stored in a cool, well ventilated area and away from flammable materials (e.g. paint).

Oxygen Gas Cylinders

These are most commonly used in the hospital setting and are prescribed for the home for getting out and about and as an emergency backup. Oxygen tanks (cylinders) contain oxygen as a gas under high pressure (usually 200bar). One litre of oxygen at 200 bars equals 200 litres of oxygen at normal atmospheric pressure (sea level). The tanks come in various volumes, from 500mL to 10L's. It is possible to know exactly how long a single full cylinder will last when operated at a fixed flow rate (worked out with readily available tables). Advantages with this delivery system are that they function independently of an electric supply and are transportable (in specially designed backpacks or trolleys). Disadvantages include the weight of the steel tanks and there is a risk that the tanks may explode if damaged (due to the high pressures involved) or near a heat source or naked flame and liquid paraffin based products therefore caution is placed when using cylinders. To have a better perspective of what this level of pressure equates to, consider that a normal car tyre is only between 2.5 to 3.0 bar (versus 200!). That is why oxygen tanks should only be securely stored in special storage devices/racks or flat on the ground. They should never be allowed to roll around or be stored in the back of your car.

How is the oxygen inhaled?

Rubber tubes of up to 8 metres in length (*long* lines; ***see*** **Figure 20**) are connected from the oxygen supply to the patient. The oxygen can then be inhaled through either the nose (nasal cannulas) or the nose and mouth simultaneously (a mask). The longer tubes provide greater freedom/range of movement for children (especially small ones) not able to move the oxygen source easily themselves.



Oxygen Nasal Prongs/Cannula

This consists of a soft plastic tube connected to the oxygen supply, which is then looped over both ears and has dual nasal prongs to deliver oxygen directly into the nose (see Figure 21). It may be necessary to tape the tubing down securely with a plaster in smaller children. The maximum flow rate of oxygen with these is 8 litres per minute, but they have the advantage of being useable while eating, drinking and being washed.

Oxygen Masks

These plastic masks enclose both the mouth and nose, and are attached with an elastic band around the back of the head. They can deliver oxygen at a rate of up to 10 litres per minute, but need to be removed for most activities (e.g. eating, drinking and facial care). Some children find these more comfortable for sleep than nasal cannulas, but they can slip off easily.

Are there adverse effects from long-term oxygen therapy?

While excessive oxygen supplementation can result in problems in prematurely born babies, this is not a danger for full-term babies or older children. There should therefore be no fear of causing harm by giving too much oxygen to your child. Continuous oxygen therapy can lead to drying out of the nasal mucosa, which may result in crusts or recurrent nasal bleeds. Should this be the case, please let your doctor know. It is possible to have a humidifier attached to the oxygen supply over 1lpm flow, which can prevent this drying out of the mucosa. Please note that oxygen suppliers do not provide this extra equipment, so you would need to discuss humidification with your child's healthcare professional. In general, humidification of the oxygen is not necessary.

How can everyday life continue with oxygen therapy?

Supplementary oxygen normally results in an improvement in your child's condition, especially their exercise tolerance. Whereas before they were too weak to go to school or nursery, oxygen therapy makes attendance a possibility. That is, rather than restricting your child, oxygen therapy normally results in more freedom for them. Dependent on the kind of oxygen supply and flow rate required, it may be necessary to set up an additional oxygen supply at school or nursery. Some parents report that having to wear nasal prongs can result in social difficulties of being stared at in public or teased at school/in nursery by other children. In the latter case, an education session at the school or nursery can be effective in stopping some of this. Please discuss these issues with your healthcare professional.

What can I do if my child is too sick to breathe?

Unfortunately, with some very sick children, oxygen therapy alone is not sufficient to maintain adequate oxygen saturations and additionally they may not be able to eliminate enough carbon dioxide from their blood. In these cases, the child will require assistance with their breathing in the form of ventilatory support (*mechanical ventilation*). There are multiple ways of providing ventilatory support. These can be roughly divided into two groups: *invasive and non-invasive ventilation*. The former group is characterised by having direct access into the lower airways with a breathing tube; either by directly through an incision in the throat (*tracheotomy*) or by guiding it down through the mouth or nose into the trachea (*intubation*).
Most children only require mechanical ventilation temporarily or intermittently (e.g. overnight or during acute deteriorations). Others may require continuous ventilation. These children require support by specially trained nursing care personnel, including while at school or nursery. Again continuous ventilation does not completely preclude normal daily life (***see* Figures 10, 23** and **24**). Nonetheless the care of children dependent on ventilation is very demanding for all involved. Fortunately, there are clinics nowadays that provide specialised additional care and support for children with these complex issues.

Figure 22:

This is Josefin. She is 10 months old. She has been diagnosed with ***congenital alveolocapillary dysplasia***. Right after birth she has been critically ill and had to be ventilated. Her condition improved and she only needed little amounts of oxygen (***also see front page***). During the winter season her condition deteriorated repeatedly. She often needed additional breathing support by a high-flow cannula. Nonetheless, as seen in the photo above, Josefin is a very fun-loving and active child that doesn't seem to be bothered by her cannulas.

What is Non-Invasive Ventilation (NIV)?

NIV is a specialised breathing support provided to patients with severe breathing difficulties via either a specifically designed over the nose and mouth mask or a nasal (nose) mask. These masks are connected to a breathing (ventilation) machine with flexible tubing and improve oxygen uptake in sick children. This improvement is achieved by being able to increase the pressures of the air in the lungs. One technique is by increasing pressure both during inhalation (the *peak inspiratory pressure, or PIP*) and the end of expiration (the *peak expiratory pressure, or PEEP*, this is a lower pressure than PIP). Another technique is called continuous positive airway pressure, or CPAP, which can be sufficient to assist a child's breathing in some cases. So called high flow nasal cannulas (***see* Figure 22**) provide a maximal flow rate of 50 litres per minute, secondarily resulting in continuous positive pressure in the lung and can function to a limited degree similarly to CPAP.

Irrespective of the method, the overall goal of NIV is to alleviate the workload of spontaneous breathing (it helps keep the lungs inflated). Initially NIV can be very uncomfortable and frightening for children, which is why practice in their use requires patience and reassurance. Once the children become familiar with NIV and have had a chance to notice that they feel better on it; they will not only tolerate it better but also ask for it themselves.

What is Invasive Ventilation?

With invasive ventilation, the breathing support is achieved by inserting a breathing tube directly into the trachea, either via a cut into the throat (*tracheotomy*) or passing it down through the mouth or nose (*intubation*). Intubation is primarily used for ventilation in the setting of acute emergencies, when non-invasive ventilation has proved inadequate in correcting low blood oxygen and/or high carbon dioxide levels. The breathing tube is very uncomfortable and poorly tolerated by conscious children, hence most children requiring ventilation via intubation need to be deeply sedated. Much higher pressures can be achieved with invasive ventilation as compared to non-invasive. When it becomes apparent that a child may require prolonged or long term ventilatory support, a small cut (*tracheotomy*) is made beneath the voice box (*larynx*) through which the breathing tube can be introduced (*tracheal cannula*) into the windpipe (*trachea*).

Which medications are used to treat chILD?

As previously stated, **chILD** is not one single medical condition. Instead it is a myriad of different lung diseases in children, which would not unexpectedly often require different treatments for each disease. Thus, the treatment should be guided by the underlying cause of each disease. Severe excessive inflammation often plays a significant role in many forms of **chILD**; hence medications that suppress inflammation are commonly used. Unfortunately, no medication is currently capable of curing any of these diseases. Rather, the medications that are used may improve the course of the disease and/or suppress symptoms (i.e. make the child feel better). Moreover, it should also be noted that these medications often have quite significant side effects. Therefore, before starting any of these medications, the doctor will for each individual case weigh up the potential benefits of the medication versus the burden of its potential side effects. As the parent it is important that you ask why your child is receiving a specific medication, what are its common side effects and what side effects do you need to watch out for. It is also critical to intermittently re-question the on-going need for any of these medications, as they generally will be given over months and not infrequently years. Frequently on-going benefit can only be demonstrated by ceasing the medication and monitoring the child for any changes in their condition.

You will find that children with the same condition frequently have completely different treatments in different specialist centres. This is due to the rarity of these diseases, resulting in the majority of them not having any clinical studies about the safe or effective use of the medications in their treatment. Hence, there are neither established treatment guidelines nor specifically approved medications for the treatment of **chILD**. Most of the medications are

well known proven treatments in other diseases, but we do not know whether they are truly beneficial or under what circumstances they are beneficial in treating **chILD**. Therefore, it may be suggested to you to engage in an active clinical study (provides a controlled setting for safely observing and monitoring their use). By actively asking your child's doctor whether the treatment of your child can and is being guided by any available clinical studies, you can help optimise their treatment plan. **Chapter 12** has further details about clinical studies into **chILD**.

Glucocorticoids, Cortisone and Steroids

Cortisone is the most commonly used medication in children with **chILD**. It is a natural messenger substance (*hormone*) and is produced in humans in the adrenal gland. Essentially it is involved in the regulation of many metabolic processes. Cortisone is changed in the body to its active form, *cortisol*. Cortisol is then itself changed into several other active substrates with similar structure but very different effects on the body. Thus cortisone is not really one substance, but a group of related substances collectively kept under the family name of Glucocorticoids (or *steroids*). The most commonly prescribed steroids are *prednisone*, *prednisolone* and *methylprednisolone*. Not only do steroids influence electrolyte balance and the metabolism of fats and sugar, but they also have a powerful anti-inflammatory effect. Steroids are not particularly toxic or harmful for a person's health, but as with many things the balance between getting the right dose and relieving symptoms is crucial. Imagine what would have to be written on a box of chocolates, if chocolate was a medication: "*Warning: excessive consumption can result in severe derangement of fat and sugar metabolism. Humans that over long periods of time consume chocolate excessively have a significantly raised risk of developing diabetes, hypertriglyceridemia, arterial hypertension, coronary disease, arteriosclerosis, heart attack, stroke .... Lethal cases that are related to chocolate consumption have been repeatedly documented*." The seriousness of side effects from steroids should not be diminished by this example; rather it should be understood that the risk of side effects is more related to the quantity taken.

What side effects can steroid therapy have?

High dose steroids in most cases are well tolerated. In general side effects tend to resolve with either reduction or cessation of the medication.

Warning: if your child has had a prolonged course of steroid therapy, the treatment must not be suddenly ceased. It must be slowly reduced over time (*tapered)*, to avoid the risk of a life threatening cortisol metabolic derangement.

Figure 23:

This is Joris. He
that it has yet to
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him around the c
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would improve a
to help maximis
infection by a
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optimal nutrition
improve week
ventilation.

Figure 23 (Continued):

Initially he remained oxygen dependent off ventilation but the tracheal cannula was left in as it made it easier for him to breath (***see* top right** and **bottom left picture**). Joris was a lively boy who continued to develop wonderfully from this point. The ***top right*** picture shows him riding in his bobby car with a very long oxygen tube connecting him to his oxygen supply. As he continued to grow his condition continued to improve. The ***bottom left picture*** shows him playing in a sandpit, at which time he did not require oxygen therapy (but the tracheal cannula remained in place). The ***bottom right picture*** is the most recent picture of him at 2 years of age. He had had the tracheal cannula removed 6 months prior to it and had only a small residual scar at its site. He no longer required supplementary oxygen, loved being active and started to walk. You can see on the picture that he still required the nasogastric feeding tube and unfortunately not all of the problems related to his condition had been resolved. Nonetheless the story of Joris helps demonstrate that children severely sick with **chILD** have not only a good chance to survive but also have fun in their life. Often these conditions can improve over time even when there are no medications available to treat or cure them.

How are glucocorticoids given?

They can be inhaled, swallowed or given intravenously. Usually glucocorticoids are administered as systemic therapy (distributed throughout the body) in children with **chILD**, either as tablets or injections. One method of systemic therapy is *pulse therapy*, in which the child receives a three-day course of once daily, high dose, quick intravenous infusions of the steroid. This is usually done as an inpatient treatment and it tends to be well tolerated, rarely having side effects. Sometimes it is necessary to give concurrent oral glucocorticoids, as "shock therapy".

The daily administration of glucocorticoids (usually more than 10mg prednisolone daily for adults and teenagers, or 5mg prednisolone for small children) is more often associated with side effects over time. Hence this type of therapy is normally restricted to a course of maximally weeks and then tapered as quickly as possible. During this time the child will frequently be given a medication to suppress acid production in the stomach to reduce stomach discomfort or pain. They may also receive vitamin D and calcium supplements to protect from osteoporosis (brittle bones).

Inhaled steroids tend to be the best tolerated with the least side effects. But unfortunately the dose they deliver is inadequate to treat **chILD**. The only time this technique tends to be used is when the child also suffers from hyperactive airways (the airways tend to spasm and narrow e.g. during infections).

When is it appropriate and not appropriate to give steroids?

It is always appropriate when the child is unwell and it is suspected that inflammation plays a major role in the development or maintenance of the disease. In this instance a therapeutic

trial of normal pulsed therapy is given. The opposite is true, when the disease is felt to be associated with malformed lungs and that inflammation does not appear to be playing a significant role. When the disease cannot be clearly classified, then a therapeutic trial of steroids can be justified.

In the UK, when receiving oral glucocorticoids (steroids) you should receive a steroid card from the Pharmacist. This provides information to you on the risks of steroids and information to provide to doctors if your child is more unwell (particularly if they are exposed to someone with Chicken Pox).

Table 6: Possible side effect of Glucocorticoids

- Suppression of the body's natural cortisol production (*see above*)
- Increased appetite
- Weight gain
- Slow gain in height
- Inflammation of the stomach lining (***Gastritis***)
- Stomach ulcers (***Peptic Ulcers***)
- Mood swings
- Stretch marks, as in pregnancy (***Striae***)
- High blood pressure (***Hypertension***)
- Elevated blood sugar levels (***Hyperglycaemia; Diabetes mellitus***)
- Brittle bones (***Osteoporosis***)
- Impairment of the immune system (***Immunosuppression***)
- Decreased or cessation of growth
- Clouding of the lens of the eye (***Cataracts***)

Hydroxychloroquine

Hydroxychloroquine is a medication that has general use in the treatment of malaria (a tropical infectious disease). It is therefore appropriate to consider it an antibiotic. Additionally, it stimulates the production of surfactant, inhibits the scarring process (*fibrosis*) of the lung architecture and has anti-inflammatory effects. Currently hydroxychloroquine is not approved for use in **child** (in fact no medicine is approved for chILD), but because of the above-mentioned properties it has been used for many years and there are many reports of children with **chILD** responding well to this therapy.

What are the side effects with Hydroxychloroquine therapy?

Most of the reported side effects from hydroxychloroquine are dose related and are only rarely seen with the usual doses used for children (6-10mg per kilogram of body weight per day). **Table 7** lists the "frequently" reported side effects with this therapy. Frequently means that

they occur in more than one per 100 treated patients. All patients requiring long-term hydroxychloroquine therapy require yearly eye examinations and regular blood tests.

Table 7: Frequent Hydroxychloroquine Side Effects

- Decreased appetite
- Moods swings (***labile***)
- Headaches
- Blurred vision
- Stomach ache, nausea
- Flatus, diarrhoea with weight loss and/or vomiting
- Skin rashes and itchiness (***pruritus***)

How is Hydroxychloroquine given in chILD?

It is given as a tablet, capsule or liquid suspension once to twice daily. Duration of therapy is usually in terms of months and improvement is normally seen after 4-8 weeks. If there is no improvement after that time, then the therapy should be ceased.

When should Hydroxychloroquine be used and when not?

In principal hydroxychloroquine can be used in every type of **chILD**. Reviewing the available published cases, it appears to work in roughly half. Unfortunately, there is still not enough published information to sufficiently predict who will benefit and who won't. Logically those children with **chILD** caused by developmental disorders or delayed development of the lung are unlikely to benefit. Again when the exact type of **chILD** cannot be determined, a trial of this treatment can be justified because it is generally very safe.

Azithromycin

Azithromycin is an antibiotic in the macrolide family, normally used for treating bacterial infections. It also has anti-inflammatory properties.

In the 1980s it was discovered that macrolides could be beneficial in people with chronic lung disease. There is a severe chronic lung disease almost exclusively diagnosed in people of Asiatic decent called ***diffuse panbronchiolitis***. Until the introduction of macrolide therapy to their treatment, most patients afflicted with this condition died. It was noted by treating doctors that those patients who had received a macrolide for an infection improved after a short while. Most even became symptom free. Subsequently, this previously often fatal condition has become a highly curable disease. Since this observation, many patients worldwide with various lung conditions have been trialled on macrolide therapy (including children with **chILD**). Beyond a few case reports of children having benefitted, there are no systematic studies demonstrating efficacy of macrolide therapy in **chILD**.

What are the side effects with Azithromycin therapy?

Continuous azithromycin therapy is well tolerated by most children with very severe side effects being very rare. Infrequently gastrointestinal symptoms like diarrhoea occur (azithromycin stimulates gut motility). As with all antibiotics, children may be allergic to them. Sometimes allergic reactions occur even after the child has had no difficulties taking the medication for a long time. The most common symptom is an itchy skin rash. When this occurs notify your doctor. Again a problem with prolonged use of any antibiotic is the risk of developing bacterial resistance to the antibiotic. Studies have also demonstrated an increased rate of detection of atypical tuberculosis in patients with lung conditions who were treated with azithromycin.

How can Azithromycin be given?

Azithromycin has a very long duration of activity in the body and therefore needs to be taken usually only three days a week (as either liquid or tablet). Benefit may not be seen until after 12 weeks of therapy, but if there is no improvement after that time then it should be stopped.

When is it appropriate and not appropriate to treat with a macrolide?

A therapeutic trial can be considered whenever a particular form of **chILD**, that is believed to be largely triggered or continued by an inflammatory process, fails to improve adequately with glucocorticoid therapy. Whenever it is felt that an inflammatory process has little role in the disease or it is primarily caused by a developmental disorder or developmental delay of the lung, then there is little justification to treat with a macrolide. Nonetheless, in those cases where the exact form of **chILD** cannot be established, a therapeutic trial can be justified because the treatment is very safe.

Are any other antibiotics used to treat chILD?

At the moment there are no other antibiotics currently used to treat **chILD**. They may be utilised in the setting of reduced immune function (*immunosuppression*), sometimes as continuous treatment. This suppression can be because of the underlying disease or secondary to medical therapy (e.g. cortisol).

Are there other medications to treat chILD?

The medications listed above represent only the most commonly used medications. Individual cases may require alternate anti-inflammatory medications to treat them, but it is beyond the scope of this handbook to go into the details of all of them.

Does every child with chILD require medications?

No, medications are not required to treat every case. For some medical conditions, medications prove to be ineffective or are completely unnecessary. This is especially true in

conditions secondary to structural changes or developmental disorders of the lung. Furthermore, children with *neuroendocrine cell hyperplasia of infancy* (NEHI) typically respond well to oxygen therapy but not to any of the above listed medications. In addition, it must be reiterated, that there are no systematic studies demonstrating the effectiveness of any of these named medications in the treatment of **chILD**.

Figure 24:

This is Henry. He suffers from ***Cantu Syndrome***, a very rare inheritable disease, which also affects the lung. It was noted right after birth that Henry had very rapid, laboured breathing. His condition acutely deteriorated, recurrently, in the setting of mundane viral infections. During these deteriorations he often became critically ill and required invasive ventilation (***top left picture***). In-between these infections he had a poor exercise tolerance and required supplementary oxygen (***top right picture***). Subsequently, he was started on pulse cortisone therapy and had a substantial improvement in his condition. Quickly he ceased to require supplementary oxygen, had a normal exercise tolerance and viral illnesses rarely resulted in any further significant deterioration. The bottom right picture shows him on summer holidays with his older brother. Mainly he now leads an almost normal life. During flu-seasons, however, he has had several episodes of deterioration that required hospital stays and partially even invasive ventilation.

The fact that there are no proven medications in the treatment of **chILD** often results in feelings of helplessness, anxiety and fearfulness. Yet comfort can be taken from the fact that frequently children will to some extent "grow out" of their illness. That means from year to year they tend to improve even without the use of medications. This is very important to know and understand, as it can save the children from pointless and potentially harmful therapies. It is critical to regularly evaluate every child on a medical therapy to see whether it is required or not. Surprisingly, stopping or not commencing a medication often requires more courage than starting one. Furthermore, it is not unusual that more benefit can be derived from ceasing a medication than continuing it or starting an additional one. The use of all medications should optimally be done in the setting of clinical studies; please actively seek this!

If nothing else helps, can lung transplantation save my child?

In very rare, severe cases a child will progressively deteriorate despite medical therapy. Subsequent lung transplantation is considered when; exercise tolerance is significantly reduced in everyday life; daily activities are only possible under significant strain (or not at all); and the progressive deterioration of the lung function leads to a concern for survival.
Accordingly, the goals of transplantation are:

1. Improvement in the quality of life,
2. A longer survival than without transplantation.

Unfortunately, in each individual case there is no guarantee of these. However, they may be easier to achieve when great care is taken with patient selection and by carefully planning the time of listing (***see*** also **Figure 10**). Furthermore, being major surgery it comes with a substantial risk for major complications, including in the worst-case scenario death (potentially during the surgery or soon after the operation). Therefore, transplantation is only ever considered after all other therapeutic options have been exhausted, and that the potential benefits of the surgery outweigh its risks. The reverse is true when the above goals are unlikely to be achieved. Additionally, there are other non-medical considerations that contribute to the likelihood of success or failure of lung transplantation. A major one is patient behaviour. Is the patient reliably able and willing to comply with life-saving therapies post transplantation? Whereas the medications for the underlying **chILD** were relatively a recommendation, the post-transplant medications are mandatory for survival. Accordingly, there is a mostly unwritten "*therapy contract*" when undergoing transplantation. It includes that the patient and their parents are obliged to fully comply with the medical management as prescribed by the transplant team. The exact adherence to this treatment plan is not only in one's own best interest, but also follows the obligation to respect the organ donor (and their family) and those other children still on the waiting list (for their transplant lung). Again it is not possible to discuss all the details involved with lung transplantation in this booklet.

Figure 25:

This is Ben Ole. He was noted at the age of 3 months to have rapid, laboured breathing and had ceased gaining weight. At the age of 4 months he was diagnosed with ***Neuroendocrine Cell Hyperplasia of Infancy (NEHI)***. The diagnosis was a great burden to his parents as they were very concerned about their son. Initially Ben Ole required continuous supplementary oxygen. He thrived splendidly on oxygen therapy, improving quickly without the addition of any medications. Subsequently he only required oxygen at nighttime or during infections. Today Ben Ole is 3 years old. He has long periods of time where does not require oxygen. During infections he still requires oxygen at times. He is lively, active and does not hold back when compared to his peers. Ben Ole is about to effectively outgrow his medical condition, even without medications.

http://www.nhs.uk/Livewell/Goodfood/Pages/eatwell-plate.aspx

Table 8: Examples of Medical Conditions requiring a Special Diet

- Food allergies (confirmed with a food provocation test)
- Fructose Malabsorbtion
- Lactose Intolerance
- Coeliac Disease
- Congenital Metabolic Disorders
- Severe Congenital Immune Deficiency
- Children post Lung Transplantation
- Children with Malignancies (Cancer)
- Children post Bone Marrow Transplantation

What do I need to consider for my child's nutrition, when their immunity is extremely suppressed?

Any child with a significantly weakened immune system, such as with *severe congenital immune deficiencies* or secondary to medication (including during chemotherapy and post-transplant anti-rejection medications), have an increased risk of infection. Importantly, this risk includes infection being transmitted by means of food. It is essential in this setting to follow these recommendations:

- Do not leave food that requires cooling at room temperature;
- Meat, processed meat (sausages etc.), seafood and dairy products should be stored in the fridge;
- Raw meat must be cooked completely through and either eaten on the day of purchase or promptly cooled for later consumption;
- Only use fresh eggs and cook them completely (i.e. egg white and yolk must not be "runny" or "soft");
- Slightly mouldy food products contain mould spores and toxins, and must be disposed of. Heating may kill the moulds but does not remove the toxins.
- Do not leave warm food out: eat it freshly cooked or store quickly (< 2 hours) in the fridge after cooking. Leftovers in the fridge need to be heated completely through before consuming.
- Follow the expiry dates on food products. Under no circumstances use them post this.
- Carefully adhere to personal hygiene in the kitchen:
    - Wash hands before and during food preparation (especially raw meat, seafood and eggs);
    - Frequently clean work surfaces;
    - Strictly separate the working equipment (cutlery, bowls, cutting board etc.) when preparing raw meat, eggs, salad etc. for other family members;
    - Wooden boards cannot be used for food preparation or as a substitute for plates (plastic, stone or porcelain boards are acceptable alternatives);

- Dishcloths, sponges, brushes and tea towels that are used to do the dishes, need to be dried and changed frequently;
- Use paper (kitchen) towels to dry food.

What can be done if my child does not gain weight?

Children with **chILD** often have an increased caloric requirement because of their increased breathing rate. Often this means that they have to eat more than healthy children. When weight gain is good, then the child is likely to be in relatively good health. On the other hand, weight loss and poor growth are serious warning signs of a significant underlying problem and need to be brought to the doctor's attention. Hence, all children with **chILD** need to have regular measurements of weight and length and the results plotted on a growth chart which ought to be shown to you at every visit. Inadequate weight gain often indicates insufficient caloric intake for the child's needs and requires the assistance of a dietician to calculate the *required daily caloric intake*. To assist with this, a *food diary* should be used to record the child's current dietary intake each day, and hence determine *their actual caloric intake*. Should the actual intake be less than the required, and the child is not able to eat enough to compensate for this deficiency, then the child will likely require *supplemental high caloric drinks*. These special drinks contain 1-1.5 kilocalories per mL, come in several different flavours (which children seem to love) and are usually on prescription through your child's General Practitioner (GP).

What can be done when my child is not able to drink or eat enough by themselves?

Some children become too weak to consume the required amount of food by themselves. This can be resolved by using a special feeding tube going into the stomach. Usually feeding by these tubes (*enteral feeding*) is needed only temporarily but sometimes they can be required permanently. Another advantage of these tubes is that some (*not* all) medications can also be given through them. Before trying to give any medications through the tube, please check with your doctor that it is safe to do so.

There are two main routes of inserting these feeding tubes: *nasogastric* and *percutaneous endoscopic gastrostomy* (*PEG*). The *nasogastric tube* is a soft, thin plastic tube, which is passed via the nose down through the food pipe (*oesophagus*) into the stomach and taped into place with a plaster onto the cheek. Although the insertion is uncomfortable, it is not painful and does not require anaesthesia (sedation). Children usually become used to the tube in a very short amount of time, and further they do not interfere with normal eating. As a side note, it is important that the feeding tubes are changed every 3 to 6 weeks, with this simple insertional procedure being easily learnt (and performed) by their parents.

The daily supplementary feeds given via the nasogastric tube can be given, either as several short duration "boluses" or continuously over several hours with a special pump. The later method is required when the child cannot tolerate large volumes of fluid in a short period of time (i.e. they complain of nausea, bloating or even vomit after each bolus). A problem with nasogastric tubes is that they can be relatively easy for a child to accidently or deliberately remove. This may result in merely the frustration of having to frequently reinsert them; to the

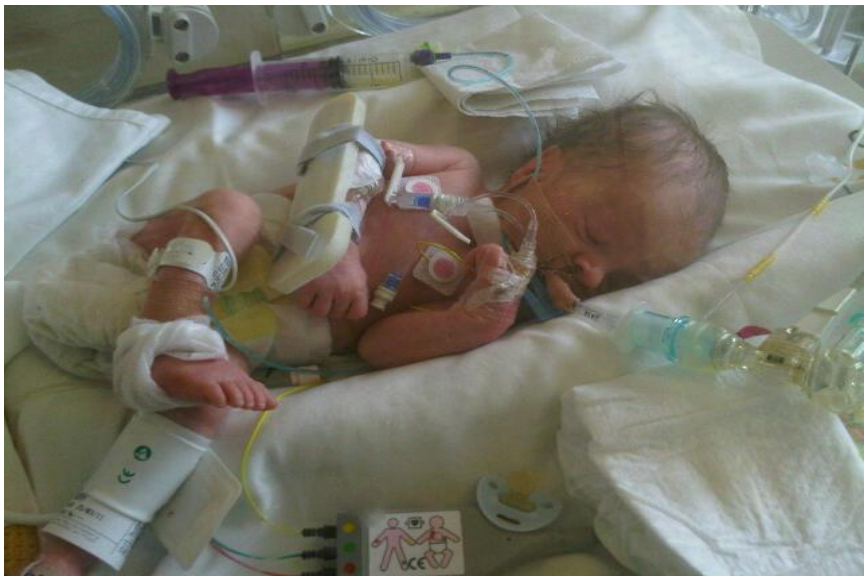
more significant problem of ***aspiration*** (swallowing food or fluid into the lungs) if the tube is pulled out during a nasogastric feed. Hence, the child should only have nasogastric feeding while being observed, which may prove problematic in the community (outside of the hospital setting). Furthermore, they can be awkward for the child when playing, potentially becoming caught on things or attracting the attention of other children (possibly resulting in uncomfortably questioning, staring or even teasing).

With regards to *PEG tubes*, they are inserted directly into the stomach through a small cut (*incision*) made into the skin on the left upper quadrant of the belly (*abdomen*). Small protective plates on either side of the incision (one skin side and the other on the surface of the inside of the stomach) help pin the tube in position and stop accidental dislodgement (***see* Figure 26**).

Figure 26: A diagram of a Percutaneous Endoscopic Gastrostomy (PEG)

1. Tube through which feeding can be given.
2. Superficial securing plate on the skin.
3. Transit of the tube through the skin into the stomach.
4. Opposing, securing plate on the interior surface of the stomach.
5. Stomach

The interior placement of the tube is done endoscopically and requires a general anaesthesia; it is usually a very safe procedure. Typically, complications may include wound infection; transient wound pain from the operation or tube blockage. Approximately 10 days after insertion (the time for the wound to heal) the on-going maintenance of the PEG becomes easy. It simply requires daily cleaning with water, drying and then gently rotating it (prevents the tube from adhering to the surrounding tissue). Since it is hidden under clothing, it remains "*out of sight and out of mind*" avoiding the attention of other children. Furthermore, children with PEGs are able to swim, bath, participate in sport, and do most other activities just like children without them. Most importantly, children with PEGs need to be encouraged to continue eating independently and not to "unlearn" normal eating. Finally, when the PEG is no longer needed, it can be taken out easily.



CHAPTER 6: RECOGNISING COMPLICATIONS

How can I recognise that my child's condition is deteriorating?

Many parents fear that they may not recognise quickly enough that their child is getting sicker. This tends to be greatest during the time soon after the diagnosis is first made. There is the fear of missing things through lack of knowledge and also of doing something "*wrong*". You do not need to be a doctor to recognise that your child is becoming sicker. Trust in yourself and your instincts, you will recognise it. It is important, especially in the initial phase, to have someone you can contact and talk with whenever you are uncertain about something. Ask who can be contacted during these times and then do not be afraid to use this help. Beyond your "gut instincts" that there is something wrong, there are several warning signs or symptoms which should prompt a visit to your child's doctor. Several of these are explained below.

Respiratory Rate

The rate of breathing, or *respiratory rate*, is a measure of the number of breathes per minute. Especially in children with **chILD**, it is a simple but effective means to assess their condition. Increased respiratory rate often indicates deterioration. But to recognise this, you first need to know what the normal rate for your child is. The best way to do this is to observe and measure your child's *baseline* breathing rate when they are relatively well. One method of doing this in children is to lay your hand onto your child's belly, and then count how often it is raised in 60 seconds (children are "*tummy breathers*", meaning that they lift their belly as they breath in). As physical exercise increases this rate and deep sleep reduces it, the optimal time to do this is when your child is awake and well rested (i.e. have not exhausted themselves just prior to measuring it). It should be noted, although there are normal respiratory rates listed for healthy children in each age group, these values will tend to be different (slower) than what will be normal for your child (their baseline rate). That is why we have not listed them here.

Shortness of Breath

Beyond an increase in respiratory rate, worsening shortness of breath (*dyspnoea*) is another indicator of deterioration. Older children from 3 to 4 years of age can reliably report being dyspnoeic themselves. In infants or toddlers you must look for the typical sign of "*indrawing*". This is where there is a sucking in (or drawing in) of the skin between the ribs (*intercostal recession*) or above the collar bones (*supraclavicular retraction*) with strained inhalation (breathing in) (***see* Figure 28**). Dyspnoeic older children will tend to sit upright and not lie down when they are feeling short of breath (sitting upright makes breathing easier).

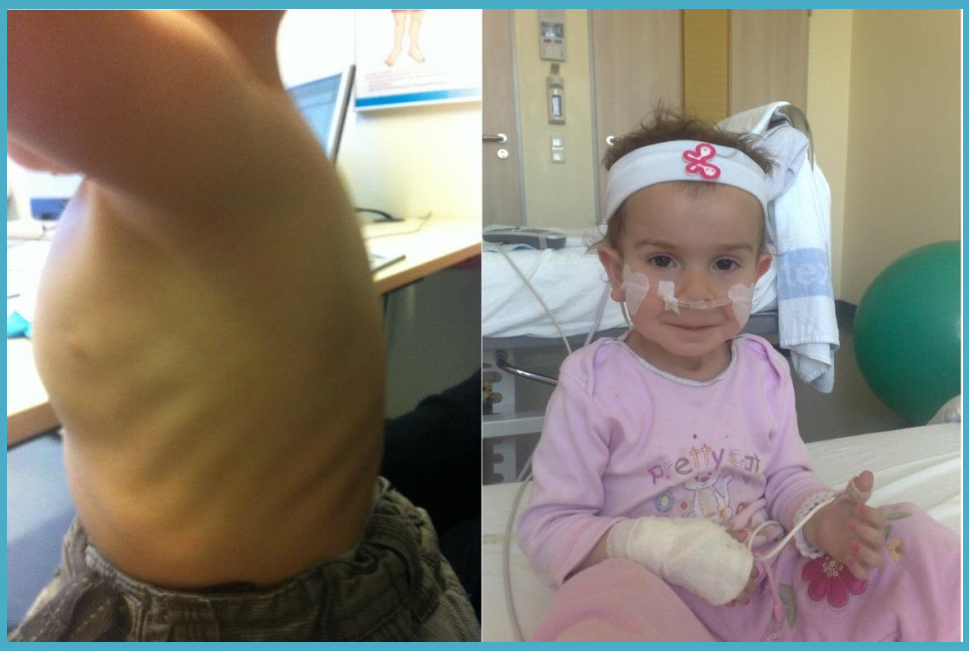


Figure 28: Signs of shortness of breath

The left picture is of a 2-year-old boy with ***pulmonary interstitial glycogenosis*** demonstrating the sign of intercostal recession or "indrawing". The right picture is of Irem (also ***see* Figure 7**) with evident supraclavicular and jugular (between the collar bones) retraction.

Drop in Oxygen Saturation

If you have an oxygen saturation monitor at home, you should understand that oxygen levels fluctuate normally throughout the day in a healthy person (lower during the night than during the day). Likewise, a person's posture can affect saturations. Some children may have lower saturations when they lie on their belly and others when they lie on their side. So as with the respiratory rate, it is important to compare saturations to a known "normal" baseline range for your child. When there is a drop of more than 5% in this baseline (or an increase in the oxygen requirement to maintain the baseline) this is an indicator of decline in lung function and that you should contact your child's doctor.

Breathing Sounds

Newly occurring or abnormal breathing sounds (e.g. wheezing or whistling sounds when breathing out) in conjunction with strained breathing (***see above*)** can be another sign of difficulty breathing. If these are persistent your doctor may provide you with an "inhaler" or "puffer" (inhaled *bronchodilating* medication) to see if there is any benefit. If there is no improvement discuss this with your doctor.

Other signs or symptoms

There is a large list (***see* Table 9**) of other potential signs or symptoms that can help indicate that your child has become sicker. Although these can also occur as a result of non-lung related diseases.

Table 9: Other Signs and Symptoms of a Deteriorating Child

- Pale skin
- Blue discolouration of the lips (*cyanosis*)
- New or worsening cough
- Not able to talk in full sentences without taking a breath in between words
- Restless, aggitated
- Listless, apathetic
- Lethargic, drowsy
- Increased sweating
- Fever
- Vomitting
- Tummy ache
- Loss of interest in eating or drinking

What can I do when my child is unwell?

Always trust your own judgement – you are the expert in your child. It can take a little while to learn how important different symptoms are – but that is why it is important to be reviewed when you are concerned. In this situation you should contact either your local paediatrician or the treating specialist centre. The urgency will naturally depend upon the severity of the complaint. Mild tummy ache, a snuffle or fever when otherwise relatively well does not suggest that your child is in immediate danger. These cases are treated no differently than for otherwise healthy children. Nevertheless, whenever you are uncertain or worried, consult your doctor. This is especially important when the underlying condition is severe, and your child is often repeatedly ill or has previously required hospital treatment for illness. With these more concerning categories it is helpful to have a plan of action established in case of emergencies, including a specific first response for any specific deterioration. Unsurprisingly, it can be very stressful during emergency situations and difficult to think clearly, so having a clear prepared plan is invaluable. Not only should this ***emergency plan*** include what specific medication is to be given, but also have a prepared list with all of the vital information for an emergency team. In addition to the list helping you provide the right information quickly and easily, it will also help third parties involved with your child's care (such as schools or nurseries) that are even less familiar with these details. This important information should include: name, age, weight, underlying diagnosis, regular medications, ventilation parameters, dosage of emergency medication given, and the names and telephone numbers of the parents and treating doctors.

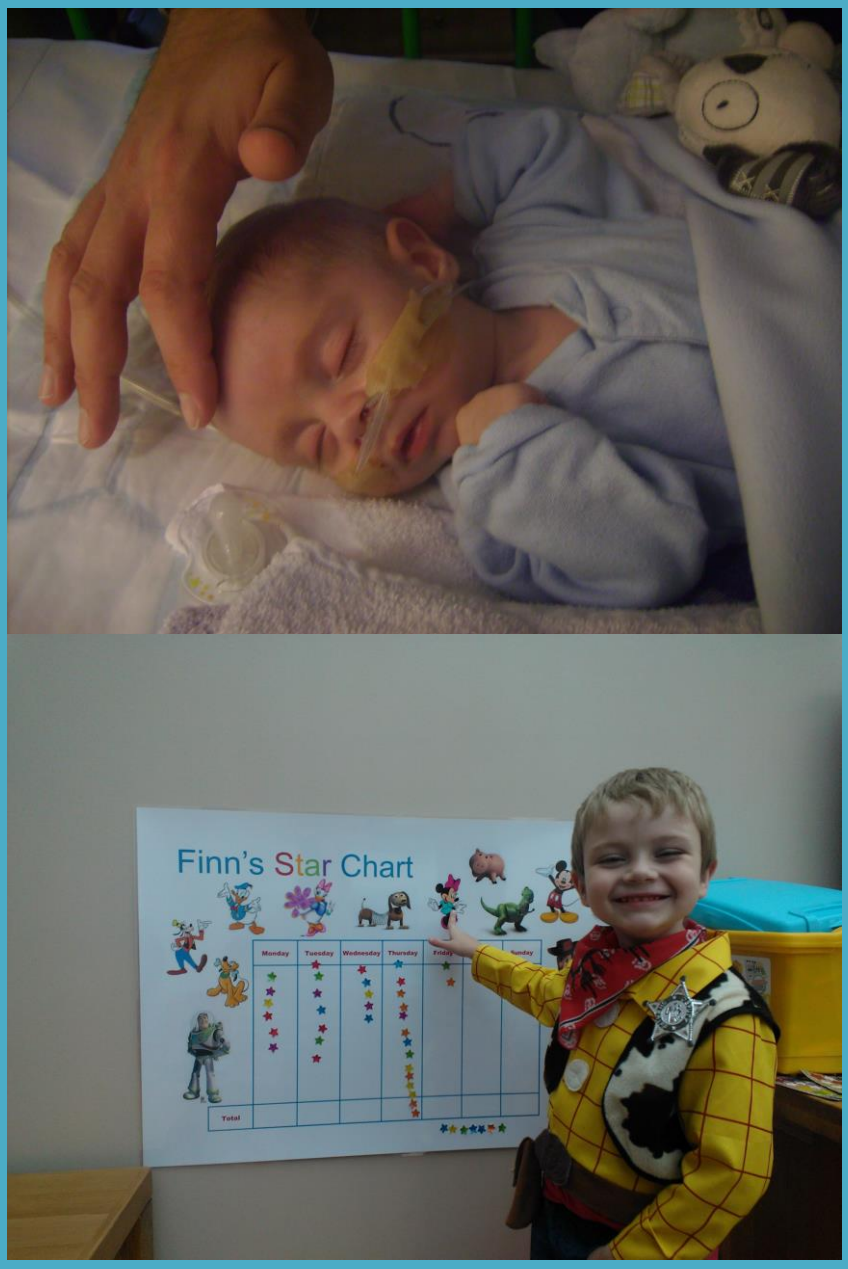


Figure 29:

This is Finn. At birth Finn suffered with a pneumothorax and was diagnosed with Protein Surfactant Deficiency ABCA3. Finn's condition was slowly deteriorating until he was diagnosed with a stage 1 Wilms Tumour (a form of kidney cancer) where the chemotherapy cleared away his lung fibrosis. As you can see, Finn is doing extremely well. Finn is nearly in 5 years remission of cancer and also only requires night time oxygen coupled with hydroxychloroquine and azithromycin medication. Finn is very happy and settled with support in mainstream school and is currently learning how to chew after many years of exclusive gastrostomy feeding.

CHAPTER 7: PREVENTION OF COMPLICATIONS

How can I protect my child from infections?

Infections are the main complication for children with **chILD**. Likewise, they are also the main reason for healthy small children to present to their doctors. This is because small children tend to have immature immune systems and physically smaller airways. Worthy of note, other than those forms caused by an immunodeficiency, children with **chILD** do NOT become ill more frequently with infections than healthy children (unless they are on steroids or other immunosuppressive medicines). Nonetheless, they do have a higher risk of becoming more severely ill. In general, one can say that, the more severe the underlying disease, the greater the likelihood of becoming severely ill with a mundane infection (most of which are caused by viruses). It should thus be decided on the basis of the relative individual risk, what infection prophylaxes may be required.

The desire to protect our children is strong but the relative risk-benefit and cost of any protective measures need to be considered. First, no measure can 100% protect your child from infection. Second, excluding your child from nursery, school, sports clubs and/or birthday parties because of the fear of exposure to infection reduces the risk, but will also significantly reduce their quality of life. Such drastic measures are only needed in individual special cases, but are by no means necessary for all children with **chILD**. The task lies with your specialist treating team to advise you which measures should be taken, after carefully weighing up all of your child's individual risks with the benefits and potential cost (often in quality of life). Blanket recommendations for all children with **chILD** cannot be given here and could not do justice to the complexity of these decisions. Nevertheless, there are some preventative measures that can be taken by all children with **chILD** and these are:

1. Implementation of all recommended standard vaccinations;
2. Yearly influenza vaccination;
3. Hand washing to reduce infection transmission;
4. A healthy vitamin rich diet;
5. Avoidance of people with a known infection.

Extreme measures, such as taking a child out of school or nursery, may be necessary in special cases. The severity of the disease and course of previous infections are key features in this kind of decision. For instance, a small child with an increasingly stable condition, clinically demonstrating only faster breathing and having had unremarkable episodes of mundane airway infections, should be able to go to nursery without any concerns. In contrast, a small child requiring oxygen therapy that has repeatedly deteriorated with infections and had multiple infections in short concession, should not attend nursery.

Are immunisations dangerous for my child?

This question can be resoundingly answered with a No. In fact, through immunisation you may be able to save your child from a potentially life-threating illness. Accordingly, your child should receive all the same recommended immunisations that healthy children should also have. Furthermore, children with **chILD** should have yearly influenza vaccinations. The only exceptions are children with a severe immunodeficiency, post bone marrow transplantation or post organ (e.g. lung) transplantation. With these children the live-attenuated vaccines (i.e. made from living functional micro-organisms with reduced disease producing ability) should not be given. Live-attenuated vaccines include measles/mumps/rubella, varicella and the new nasal influenza vaccine (***see* Table 10**). Also all close relatives with frequent contact to your child should also receive the recommended immunisations. This includes: the parents, siblings and potentially grandparents. Children with severe disease will often additionally receive seasonal vaccines (e.g. vaccines for the *Respiratory Syncytial Virus, or RSV*, are given in the first two years of life, every 4 weeks during winter). For more information please check out the latest UK vaccine schedule: **http://www.nhs.uk/conditions/vaccinations/pages/childhood-vaccination-schedule.aspx**

Table 10: Vaccination Checklist

Frequency	Vaccination
Yearly each Autumn	Influenza Vaccine (*Patient and Family Members*)
Every 5 years	Pneumococcal Vaccine
Every 10 years	Booster Immunisations: *Diphtheria; Polio; Pertussis* (whooping cough)*; Haemophilus Influenza type B* (HiB)*; & Hepatitis B*
Never in Children with Immunodeficiencies	**Live-attenuated Vaccines: *Measles, Mumps, Rubella; Varicella; BCG* and *some Travel Immunisations***

Smoking

Smoking causes significant lung damage. Therefore, there should be no smoking in the presence of children with chronic lung disease (it is particularly dangerous near children using oxygen). Furthermore, there should never be smoking in places where the child spends significant amounts of time, even when the child is not there. Providing a safer living environment for your child is even more successful when smoking parents quit. So do not hesitate to seek professional help with quitting (e.g. your GP), as they can help you find the right programme. Also consider that there are significant demands and restrictive behaviours required of your child with managing this disease. Should it be unreasonable for them to see similarly necessary changes in their parents' lifestyle? By quitting you help provide a good

role model for your child.

CHAPTER 8: NURSERY, SCHOOL AND RECREATION

Can my child attend nursery or school?

Not only is it possible for most children with **chILD**, but it should also be seen as essential. The exceptions for this occur when the child is either acutely too unwell to go (as for children without **chILD**), or if they have frequently and severely fallen ill with airway infections in the past. It is essential that the carers or teachers are informed about your child's condition and that they know what to do in case of an emergency. This includes in some cases providing a written emergency plan and ensuring that potentially required emergency medications are stored at the nursery or school. Despite these measures, a carer or teacher may refuse or be too afraid to administer emergency medications. In most cases this can be resolved as your child's specialist treating team can provide information or an education session at the school or nursery.

Another issue to be aware of is that your child may require lots of sick days off from school, with the corresponding amount of catch up homework being extremely stressful. Your child has access to special rights as they have special requirements. You should make use of these rights to help broaden the options of your child's educational care. There are several options available should your child be having significant difficulties with their schoolwork. One option includes the possibility of creating an individual education plan for your child with their teachers and headmaster. For chronically ill children there is also the option of having a specially trained school carer to accompany and support them at school. During school hours the support carer will assist your child with keeping to the school schedule, getting to classes, providing any required medical care and giving general assistance as required. The cost of these carers is provided by social services. Optimally, you should submit an application to your local social services office before enrolment or the start of a new school year, if your child requires one of these carers. Furthermore, it is helpful with these applications to include a certificate from your doctor and a letter from the school.

ChILD and sport, is it possible?

All children have the natural urge to move. This is no different for children with **chILD**. Moreover, this urge should be encouraged and NOT restricted. Indeed, the benefit of sport activities goes beyond their important social aspects, they also have a positive impact on the general health of the lung and overall health. It is only in rare circumstances that a certificate for exemption from sport is necessary. Even severely diseased children should be encouraged to participate in sport, but they should be exempted from grading and allowed to rest whenever they wish to. The fear that a child will over exert themselves is in most cases unfounded, as they will instinctively stick to their limits. Here it is more important that others do not attempt to push them past their physical limits, to become too active or too competitive. In fact, this behaviour is counterproductive, usually resulting in loss of joy and

increasing frustration as the child attempts to please others, but is not able to live up to expectations. Finally, there is no special kind of sport that is more suitable or better for children with **chILD**. The most important point with sport is for your child to have fun and remain active. Talk to sports teachers about your child getting as much exercise as they are capable.

Figure 30:

This is Matty who was born full term but within a few weeks started to get recurrent respiratory infections. Matty has been on using oxygen therapy since 4 months old and after a few hospital admissions, testing and eventually a lung biopsy, he was diagnosed with Follicular Bronchiolitis (FB). Matty also has been diagnosed with Mannon-binding lectin (MBL) deficiency, a genetic condition that affects the immune system. FB and MBL deficiency does not stop Matty being an active, happy boy who is doing very well at the moment. Matty will be starting a special needs school shortly and we wish him the best of luck.

CHAPTER 9: HOLIDAYS AND TRAVEL

Is it possible to have holidays or travel with chILD?

Yes, if the overall health of your child is stable. When oxygen therapy is not required, then the holiday planning is more simple, but in all cases chat with your medical team about how best to stay well on holiday and what to do if your child is less well. With more severely afflicted children, a good holiday plan is a necessity. In the next few subheadings we will provide several important details that need to be considered with this.

What do I have to consider in planning a holiday?

In some cases, planning a holiday with a child that has **chILD** can be very challenging. Long car trips, train rides or flights must be precisely thought through and organised. As a general recommendation, we would advise you to select a destination and the method of travel based upon your child's needs. In other words, the more severe your child's lung disease is, then the higher quality of medical care needs to be readily available at the destination. Likewise, the presence of pulmonary artery hypertension will likely preclude flight as an option (check with your doctor *before* booking tickets).

Remember that the doctors at your destination will be unfamiliar with your child and many have no or limited knowledge regarding **chILD**. Hence, it is essential to have a medical travel folder for the trip, containing copies of all the important medical documents and a list of the current medical therapies for your child.

Where can I get oxygen for my child whilst abroad?

Currently there is no provision for UK oxygen suppliers to provide oxygen for travelling abroad. Therefore, you may have to budget for this additional cost. For holidays abroad some families purchase their own airline approved portable oxygen concentrator or alternatively rent from oxygen rental companies such as Pure O2 (**http://www.healthoxygen.com**) or Oxygen Worldwide® (**www.oxygenworldwide.com**). Some charities may help with this additional cost, especially if you are in receipt of certain benefits or have a low family income. Alternatively, ChILD Lung Foundation offers this free of charge service to registered chILD families but the service is very much demand so you may have to check availability in advance.

Travel immunisations

Make sure that you contact your doctor well in advance to any planned holiday overseas. This way you can check what your child's current immunisation status is, whether any additional vaccinations may be required and still have the time to get them.

What is important to know about health insurance while overseas?

Medical care in a foreign country can be very expensive, as it is not usually covered by standard health insurance. Therefore, it is always sensible to take out additional travel health insurance when planning a holiday overseas. Pay attention to whether or not transport back home is included in this additional cover. Furthermore, watch out for exclusions of treatment costs with deteriorations in known pre-existing medical conditions. Whenever there is doubt, the best course of action is to clarify directly with the insurance company that the planned policy will cover the costs of any emergencies, despite your child having a known interstitial lung disease. Should it prove impossible to take out travel insurance because of your child's underlying lung condition, discuss things with your usual health insurer. Some of these health insurance companies will provide partial cover for medical costs while travelling abroad that would have arisen in your home country. Another option to provide you with some peace of mind when abroad is to apply for a European Health Insurance Card (EHIC). This card may help with medical emergencies and country specific state care in European Economic Area (EEA) countries. Please note that this card is not a replacement for medical insurance. Please visit the NHS Choices link for more information and the application process: **http://www.nhs.uk/NHSEngland/Healthcareabroad/EHIC/Pages/about-the-ehic.aspx**

Will I have problems with customs because of the medications?

When travelling, it is always recommended to carry your medications in your hand luggage. This way they remain easily accessible in case of an emergency and it will also diminish the risk of accidental loss. Nonetheless, different medications are restricted in different countries, and hence you will require a medical certificate when entering these countries. Check with your airline prior to departure to find out which medications you will require a certificate for. Finally, the best approach to help avoid difficulties at customs is to declare every medication with their correct quantity, and all the medical equipment you are travelling with. Your doctor should provide you with a letter outlining your medicines and any medical devices that you need to take with you on the aircraft (oxygen, ventilators etc.).

Electricity supply overseas

Make sure you know what electricity supply is available at your destination prior to leaving, especially if your child requires a ventilator, oxygen concentrator or other medical equipment requiring electricity. You may need to obtain a special power adapter for your holiday country.

What do I have to consider with air travel?

Even if your child normally does not require supplementary oxygen, you must be prepared that your child may require it with a flight. This is because at 3000m of elevation there is a

lower concentration of oxygen than at sea level (c15% rather than 21%). Children with chronic lung disease may not be able to compensate for this lower concentration, resulting in their oxygen saturations dropping significantly during the flight. It is therefore important to discuss this risk with your doctor prior to flying. For this same reason, airlines may demand a doctor's certificate declaring your child "fit for air flight". A fit to fly test could be organised by your doctor and simulates the inflight oxygen concentration (15%).

When oxygen is required for the flight, it is important to let the airline know well in advance, clarify if there is oxygen available on board and what other forms your child's doctor will need to complete prior to the flight. Since every airline deals with this issue differently, we have included a list of commonly used airlines with important information regarding this. Most airlines will not allow people to bring on their own pressurised tanks, but will allow carry on oxygen concentrators. Usually, this will require the reservation of an additional seat, which in some airlines can be without additional charge if you possess a disabled person's pass. Although every aircraft carries oxygen on board, this is limited and only intended for medical emergencies. Consequently, if your child requires the use of this oxygen, the pilot may out of necessity have to divert the aircraft to a nearby airport and call for medical assistance for your child. Further and more detailed information about the oxygen policies for most Airlines in Europe can be found at the European Lung Foundation site: **http://www.europeanlung.org/en/lung-disease-and-information/air-travel/airline-index/.**

Figure 31:

Stephanie from birth gained little weight and was diagnosed with 'failure to thrive'. After a hospital admission from a virus, Stephanie was diagnosed via a CT scan with with probable NEHI. Since Stephanie is doing very well, a lung biopsy is not required however she does require continuous oxygen therapy. Stephanie leads a full and active life and never stops smiling.

CHAPTER 10: SOCIAL AND FINANCIAL SUPPORT

If your child has a disability, financial and practical support from the Government may be available to help you manage. As soon as you think you are eligible, please make a claim as these benefits are not automatic or linked to your child's medical team.

Disability Living Allowance

Disability Living Allowance (DLA) for children may help with the additional cost of looking after a disabled child. This benefit is for children under the age of 16 and who have difficulty walking or additional needs compared to a child of their own age who does not have a disability.

There are two elements to Disability Living Allowance (DLA).

Care Component: this is for people from age three months to 64 years who have need additional care due to their disability. The financial support for this component comes in award rates lower, middle and higher.

Mobility Component: this is for people from age three years to 64 years who have mobility problems due to their disability. The financial support for this component comes in award rates lower and higher.

For more information on DLA visit: **https://www.gov.uk/disability-living-allowance-children/overview**

Here you will be able to find out the current rates of benefits, eligibility and link to online application.

Personal Independence Payment

Between April 2013 and October 2017, new DLA claims for people over the age of 16 has been replaced by the Personal Independence Payment (PIP). Children under the age of 16 who are already claiming DLA are not affected by these changes, however on their 16th birthday they will be invited to claim for PIP instead.

For more information on PIP visit: **https://www.gov.uk/pip/overview**

Here you will be able to find out the current rates of benefits, eligibility and link to online application.

Income Support

This benefit is for parents on low income who can't work because of either their own are caring for a disabled child. Income Support will be gradually replaced with Universal Credit between April 2013 and October 2017. You can make a claim through your local Job Centre Plus or Jobs and Benefits Office.

For more information visit: **https://www.gov.uk/income-support/overview**

Child Tax Credit

Child Tax Credit (CTC) is an income dependent benefit for anyone with responsibility for a child who normally lives with them. CTC is made up of elements with the basic being the 'family element'. Depending on your circumstances additional elements can be added to your claim (e.g. disability element) so keep HM Revenue & Customs notified of any family changes as soon as possible. Between April 2013 and October 2017 CTC will be gradually replaced with Universal Credit.

For more information visit: **https://www.gov.uk/child-tax-credit/overview**

Carers Allowance

Carer's Allowance is for people who spend at least 35 hours a week caring for a child who gets the middle or higher rate care component of Disability Living Allowance or either rate of the daily living component of Personal Independence Payment. There are certain rules in order to be eligible such as your weekly income and how many hours you work employed, self-employed or as a student. Carer's Allowance is also a taxable benefit so it may affect any other benefits you receive.

For more information visit: **https://www.gov.uk/carers-allowance/overview**

Housing Benefit

If you live in a rent paying household and on a low income, some or all of your rent may be covered if someone in your household has a disability. Eligibility and Housing Benefit will be gradually replaced with Universal Credit between April 2013 and October 2017.

For more information visit: **https://www.gov.uk/housing-benefit/overview**

Cold Weather Payment

For families who are in receipt of certain benefits and either you or your child has a disability, you may be eligible to get a Cold Weather Payment. Usually there is no need to apply as it will be paid automatically to you. If in doubt or to check eligibility you can check the link below or alternatively contact your energy supplier.

For more information visit: **https://www.gov.uk/cold-weather-payment/overview**

Motability Scheme

For children or young adults in receipt of the higher rate mobility element of Disability Living Allowance or Personal Independence Payment, the motability scheme can help provide a car, motorised wheelchair or scooter.

For more information visit: **https://www.motability.co.uk**

Blue Badge Scheme

Disabled drivers or people who provide transport for a disabled person can apply for this parking scheme. This enables the disabled person or carer to park closer to destinations, use larger parking bays for mobility or in some cases offer free of charge parking. Charges and entitlement rules for the Blue Badge scheme vary across the UK.

For more information visit: **https://www.gov.uk/blue-badge-scheme-information-council**

Here you can find your local council, apply online and check eligibility.

Need help with disability benefits?

The Citizen's Advice Bureau estimate that more than £16 billion in means-tested benefits and tax credits goes unclaimed every year. Even if you're not certain about your eligibility, you still can make a claim (and may be pleasantly surprised!). Disability benefits are there to help you and your family and figuring them out and filling in the forms can often be overwhelming and complicated. Fortunately, there are free expert help and advice services for you to navigate through this system.

Disability Benefits Centre (here there are different helpline numbers for each disability unit): **https://www.gov.uk/disability-benefits-helpline**

Contact a Family (an information, advice and support service for families with a disabled child): **http://www.cafamily.org.uk/**

Are there any care or respite services I can access for my child?

Caring for a disabled child can be very rewarding but also at times it can also be challenging. As a carer you may feel that sometimes you, your child or even your family may need a short break away. A coffee with a friend, a walk in nature, or even a day or over night trip can help with recharging depleted batteries. Unfortunately, there are no set services across the UK to help with respite. However, there may be services available in your local area through Social Services or any local charities. If you feel you would benefit from this type of service, please discuss your feelings with your child's GP or Pediatrician. They are there to help, signpost or even refer you or your child to local care or respite services if they are available in your area.

CHAPTER 11: RESEARCH

Why is research into rare lung diseases so important?

It may seem inefficient to spend time and money investigating individual rare diseases. But often, as in **chILD**, the sum of all the individual cases within a family or cluster of diseases results in a far more significant population. Being able to identify the underlying cause (*or mechanism*) of a disease, even if it is through inheritance, allows development of targeted treatments, which in turn can often be applied to other diseases with similar mechanisms. Hence, research into even rare diseases can be very fruitful, often providing benefits for many people.

An additional benefit with research, beyond gaining more targeted treatments, is that it increases recognition of a disease by doctors in the community. That is, the more information disseminated regarding a disease (especially its natural progression), the faster the rate and accuracy of the diagnosis of a disease. Establishing a *patient registry* (data collected from as many patients as possible, optimally from multiple centres), especially for diseases like **chILD**, is a very effective method of collating this information.

Clearly faster, correct diagnoses and targeted treatments (for potentially several diseases) are outcomes desired by everyone. This is especially true in **chILD**, where the initial diagnosis is often a long arduous journey characterised by false diagnoses (such as asthma) and ineffective (or non-existent) treatments.

Where do I find information about the current research results?

It is possible to find information regarding current and new research in the **chILD-**EU project website: **http://www.klinikum.uni-muenchen.de/Child-EU/en/index.html**

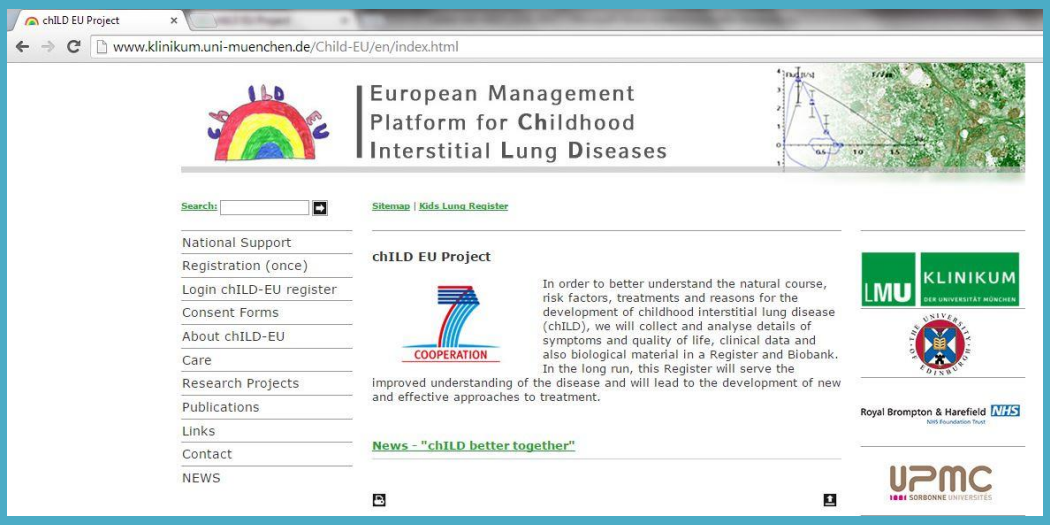


Figure 32: Webpage from the **chILD-EU Project**

How is it possible to successfully carryout long-term research into the course of such a scattered rare group of diseases?

A web-based databank (*patient registry)*, the **chILD-EU Register**, is being used to store and collate information regarding the diseases' courses from multiple centres. Naturally the data can only be submitted after gaining the permission of the parents, and does not include any patient identifiable information. What it does include is the details of your child´s symptoms and quality of life, clinical data, and results of routine medical investigations. Another feature is a Biobank, where biological material previously collected from your child during routine testing is centrally stored.

An additional benefit of this process is the inclusion of quality control for each individual case submitted, by a panel of independent experts. With this the treating doctor's diagnosis is re-examined and confirmed by the experts. The Biobank samples may be use during this re-examination. This panel may also offer expert advice for the treating team regarding the diagnosis and its management.

More details of this process are contained within the consent form you would be required to read and sign prior to the submission of your child's data. Taking part is voluntary, and you can withdraw from the study at any time, without giving a reason if you wish. Your child's treatment would not be affected by study withdrawal.

(http://www.klinikum.uni-muenchen.de/Child-EU/download/en/i_have_a_case/national-support/germany/InformedConsentCaregivers.pdf)

What will be the benefits for my child participating in the EU-chILD Register?

Beyond the initial benefit of a confirmation of diagnosis, there is unlikely to be any direct benefits for your child early on in this study. But should new insights be gained into the treatment of your child's disease during the course of the research, these findings would be shared with you. Not unexpectedly, the process of informing you would be fairly complicated, especially in view of the separation of medical data from identification data. Furthermore, the information needs to first pass approval through a local ethics committee and then subsequently would be shared with you via your child's doctor. Likewise, if you have expressed interest in being notified about involvement into any other potential clinical studies for your child, you will be contacted through your doctor when your child fulfils the relevant selection criteria.

A side benefit of this database research is that it enables the early quantification and comparison of the health care costs (including treatment costs) across Europe. For you, this helps with establishing a baseline costing standard, which may improve financial remuneration from health insurers (often a problem with rare diseases).

What clinical trials are currently under way?

Unfortunately, as described in **Chapter 4**, all the medications currently being used in **chILD** are being used "*off-label*" (being used outside of established proven or approved use). This is as a result of there being insufficient clinical evidence for their specific use in treating these rare diseases.

For this very reason, the **chILD**-EU Project has initiated clinical studies into two of the most commonly used medications, Hydroxychloroquine and Prednisolone. Specifically, to substantiate when and in which types of **chILD** they are beneficial, based upon empirical recording of the actual benefits and side effects in these diseases. Both of these studies have been approved through stringent reviews by the relevant ethics committee and the national bureau *BfArM* (*"Bundesinstitut für Arzneimittel und Medizinprodukte"* or The Federal Institute for Drugs and Medical Devices).

Clinical Trials with Hydroxychloroquine

Almost all children with severe interstitial lung disease, including those of unknown causes or genetic surfactant disorders, will undergo treatment with hydroxychloroquine. Unfortunately, this tends to be done very haphazardly currently, since the optimal timing, dose, duration of treatment, side effects and what types of **chILD** will respond are complete unknowns.

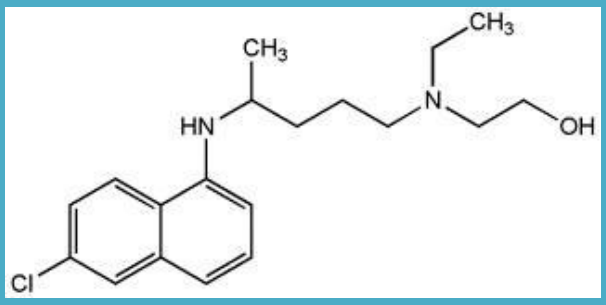


Figure 33: Hydroxychloroquine (Quensyl)

Therefore, optimally the usage of hydroxychloroquine in children should be restricted to clinical studies. To potentiate this usage under more controlled conditions, two studies have been developed whose parameters closely reflect the current clinical reality and will not obstruct on-going treatment in children.

Specifically, these two studies are trying to help with determining when to initiate therapy (***see figure* 34**) and when to cease (***see figure* 35**) with randomised placebo control. That means patients are randomly divided into one group receiving the active medication and the other a "mock" tablet (same appearance but no active ingredient).

With the initiation trial, both groups are treated with hydroxychloroquine, but the placebo group is delayed by one month (so there should be no fear of your child not being treated). Whereas the cessation trial compares in two groups, the effects of either continuing hydroxychloroquine therapy for 3 more months or withdrawing it, after having had minimally 3 months of prior treatment. Again the children are randomly assigned groups, with neither the doctor nor parents knowing whether the on-going tablet is hydroxychloroquine or a placebo (the trial is "blinded").

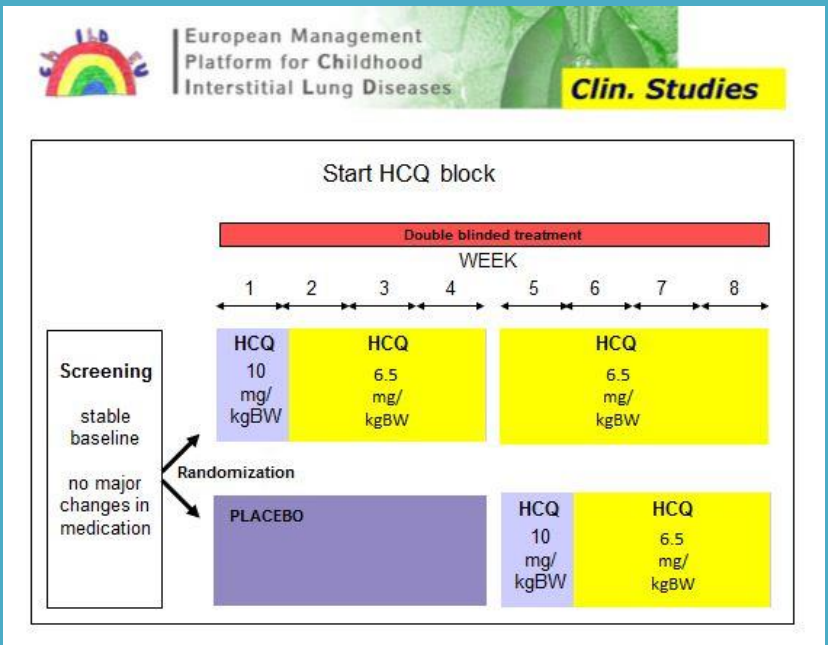


Figure 34: Initiation of Hydroxychloroquine in children with Interstitial Lung Disease

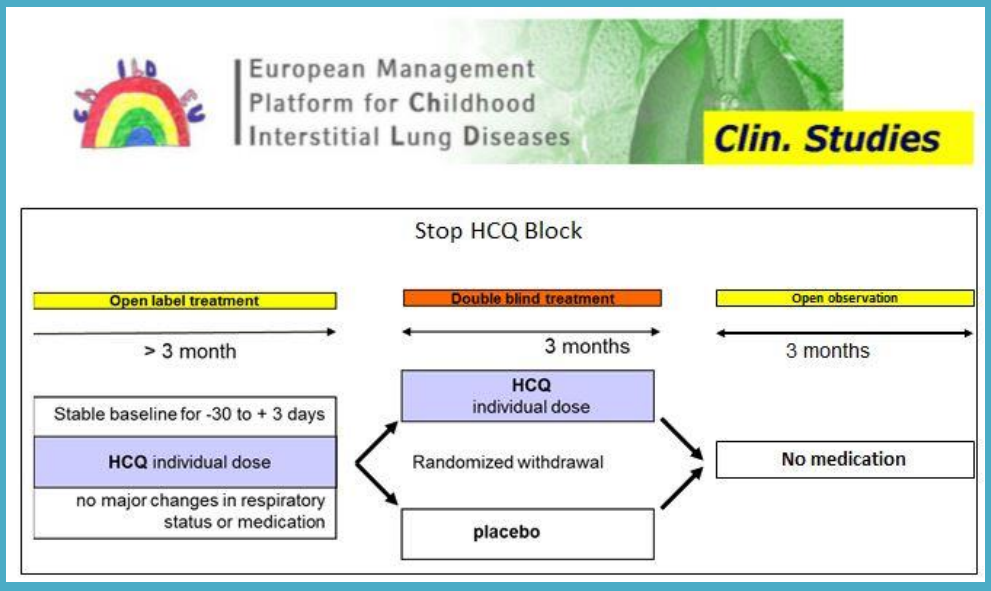


Figure 35: Withdrawal of Hydroxychloroquine in children being treated for Interstitial Lung Disease

Stop EAA Study

EAA, or *Exogenous Allergic Alveolitis*, is a less rare form of **chILD**, which may have an episodic (recurrent) or a chronic complicated disease course. Both courses can have potentially long term sequelae, and in some cases this disease can deteriorate to the point of end-staged lung disease, necessitating lung transplantation. As a consequence, children with EAA are sometimes treated with protracted courses of high dose steroids. Currently it is only possible to predict whether they will have a chronic or temporary course through hindsight. Hence, there is immense value in having a prospective, long-term observational study to help identify those features suggestive of these more malignant types and the role of steroids in affecting the course of the disease (***see* Figure 36**). Children with a new diagnosis of EAA in centres recruiting to the study will be eligible to take part.

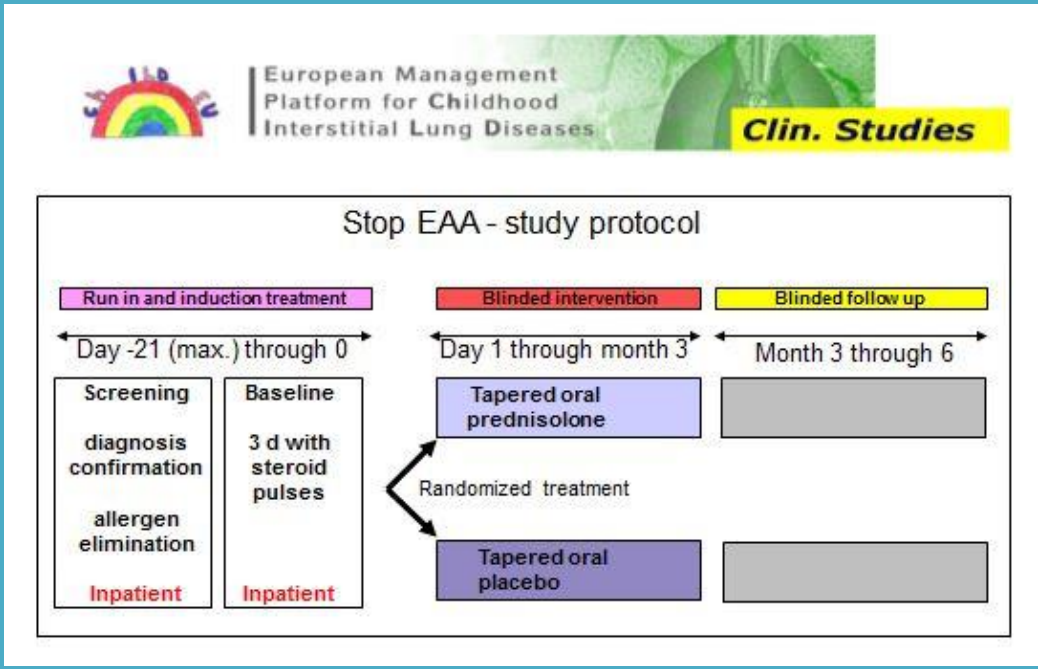


Figure 36: Study to evaluate the effectiveness of Prednisolone in the treatment of Exogenous Allergic Alveolitis

As you would expect, the more cases involved with any study (particularly of rare diseases), the stronger the findings will be for subsequently developing better care plans and targeted treatments. Hence the involvement of as many paediatric pulmonology outpatient clinics and centres as possible, with patient recruitment, plays an essential role in this research.

CHAPTER 12: SELF-HELP GROUPS, CONTACTS AND HELPFUL LINKS

Are there any self-help groups?

In the UK, there are several organisations that support **chILD** families.
ChILD Lung Foundation (http://childlungfoundation.org) is a charity that supports all **chILD** diseases and has a range of services, including providing practical and emotional support. Other organisations include the British Lung Foundation (http://blf.org.uk) who have an interest in all aspects of children's respiratory health, not just **chILD**; also there is Breathtakers OB Trust (https://www.breathtakers.org.uk) who support those specifically affected by Bronchiolitis Obliterans, and Breathe On UK (http://www.breatheon.org.uk/) who support families and carers of young people on mechanical or long term ventilation. There are many other sources of information on the internet, but often they have poor quality information and that can be incorrect. Please print or save any web information that has caused you concern for discussion with your medical team.

The Kid's Lung Register

It is hoped that the **chILD**-EU Project will provide a catalyst for the establishment of new self-help groups, which will advocate the needs of children with interstitial lung disease. For that, the register provides an easy communication platform for parents, by way of the register's forums. Consequently, to get this part of the project under way, we require your help! The website for the Kid's Lung Register has a pivotal role in the exchange of information and support (***see* Figure 37**). The registry is a non-profit organisation that has set itself the goal of promoting research into rare children's lung diseases. It is the established foundation for a variety of scientific projects.

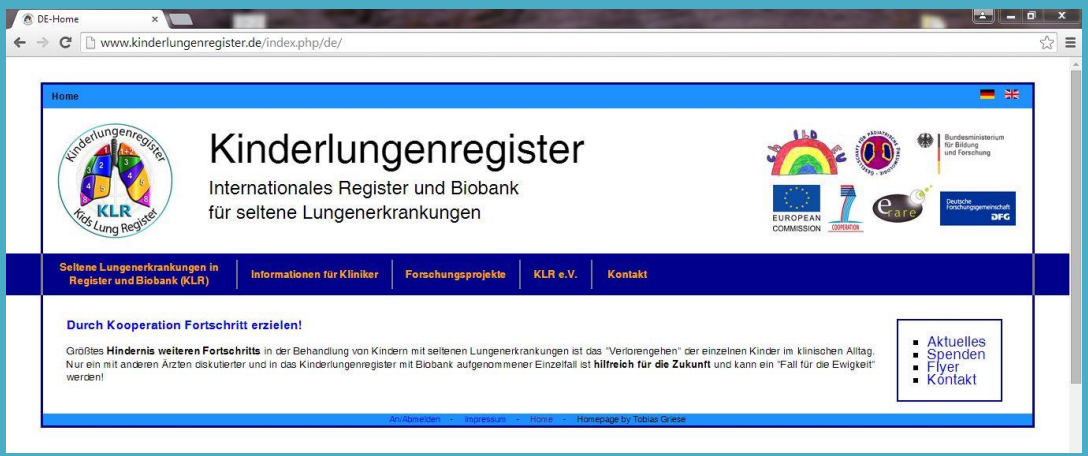


Figure 37: German webpage of the Kid's Lung Register

Helpful Internet Addresses

ChILD EU

Homepage of the chILD EU-Project.

http://www.klinikum.uni-muenchen.de/Child-EU/en/index.html

ChILD Lung Foundation

British homepage of the chILD Lung Foundation.

http://childlungfoundation.org/

ChILD Foundation US

American homepage of the chILD Foundation.

http://www.child-foundation.com

Kids Lung Register

Homepage of the Kid's Lung Register with information for patients and an international registry of rare lung diseases for medical practitioners.

http://www.kinderlungenregister.de/index.php?lang=en

Starke Lunge

German homepage for patient organisation for rare lung disease

http://www.starkelunge.de

Pequenos Pulmones

Spanish homepage of a chILD related organisation.

http://www.pequenospulmones.org/

ChILD Chit Chat

A forum for International families affected by chILD. Moderated by chILD Foundation US and ChILD Lung Foundation.

http://childfamilies.forumchitchat.com/

Organisation of Oxygen Supplies with several Airlines in Europe

Table 11: Oxygen Supplies with several Airlines in Europe

Airline	Information
Aer Lingus	Taking along your own oxygen tanks on board is not permitted. Certain oxygen concentrators can be carried however, because there is no power supply on board, you must carry a sufficient number of batteries in order to bridge the duration of the flight. Oxygen can be provided for a fee. The request must be submitted at least 48 hours before departure. A medical certificate for airworthiness is necessary. For more information please visit: http://www.aerlingus.com/help/help/specialassistance/
Air Berlin	Transportation of own oxygen provision is permitted. The container may weigh up to 5kg and must be individually protected. Valves and the containers must be protected from damage. Liquid oxygen is not allowed. A medical certificate for airworthiness is required. An application must be made no later than 7 days prior to departure. For more information please visit: http://www.airberlin.com/en-GB/site/start.php
Air France	Oxygen can be provided for a fee however, the request must be submitted at least 48 hours before departure. For a demand of oxygen more than 2 lpm, a medical certificate of airworthiness is required. Oxygen bottles can be transported under certain conditions (max. 65 cm high, 5 kg and secured against the escape of oxygen). Oxygen concentrators can be carried however, since there is no power supply on board, you must carry a sufficient amount of batteries for flight duration. For information please visit: http://www.airfrance.co.uk/GB/en/common/guidevoyageur/assistance/pmr_reservation_airfrance.htm
American Airlines	American Airlines do not provide onboard oxygen. Transportation of oxygen tanks are not permitted however, certain oxygen concentrators may be carried. Notification must be made at least 48 hours before departure. A medical certificate for airworthiness is required plus confirmation that the oxygen is prescribed. For more information, please visit their special assistance pages at: https://www.americanairlines.co.uk/i18n/travelInformation/specialAssistance.jsp?locale=en_GB

Table 11: Oxygen Supplies with several Airlines in Europe (Continued)

Airline	Information
British Airways	British Airways provide oxygen (on demand) available on board. A medical certificate for airworthiness is required. Requests must be submitted at least 48 hours before departure. FAA approved concentrators may be carried when the criteria are met for hand luggage. http://www.britishairways.com/en-gb/information/special-assistance/medical-conditions
EasyJet	EasyJet does not provide onboard oxygen however, taking along your own oxygen on board is possible (small pressure oxygen cylinders, pneumatic cylinders, battery-operated oxygen concentrators, max 2 per person.) The equipment must apply to criteria for hand luggage: max. 50 cm length, 25 cm in diameter. A medical certificate for airworthiness is required. http://www.easyjet.com/en/help/preparing-to-fly/medicine-medical-equipment
Germanwings	German Wings offers onboard oxygen only in emergencies. Taking along your own oxygen is not possible. For further information please visit: https://www.germanwings.com/en.html
KLM	Portable oxygen concentrators are allowed on board however, since there is no power supply on board, you must carry a sufficient number of batteries for the duration of the flight. Requests must be submitted at least 48 hours before departure and a medical form must be completed by a doctor. http://www.klm.com/travel/de_en/prepare_for_travel/travel_planning/physically_challenged/klm_cares.htm
Lufthansa	Taking along your own oxygen on board is not permitted. There is an oxygen supply on board, however this is limited. You should apply for oxygen provision at least 48 hours before departure. For more information visit: http://www.lufthansa.com/uk/en/Homepage
Ryan Air	Ryan Air provides onboard oxygen. An oxygen demand request must be completed on the day of booking, as the service is limited to one guest per flight. Passengers who require oxygen supply must provide written certificate from their physician, certifying airworthiness. Certain oxygen concentrators can be used on board. Please visit: https://www.ryanair.com/en/terms-and-conditions/regulations-reducedmobility/

Table 11: Oxygen Supplies with several Airlines in Europe (Continued)

Airline	Information
TUI Fly	TUIfly does not provide onboard oxygen however, transportation of own oxygen is permitted. 72 hours notice of oxygen provision before departure is required. You can take your own 2-litre bottles (UN-1072) free on board. Liquid oxygen systems are forbidden. Portable oxygen concentrators are permitted. A medical certificate is required for airworthiness. Please visit: https://www.tuifly.com/en/index.html
Turkish Airlines	Turkish Airlines provides onboard oxygen at passengers request. Oxygen tanks if carried must be empty. A medical certificate for airworthiness plus confirmation that the oxygen is prescribed is required. Requests must be submitted at least 24 hours before departure. The use of FAA approved concentrators is available upon request. Please visit for more information: http://www.turkishairlines.com/en-int/travel-information/baggage/transport-of-special-equipment
United Airlines	Oxygen tanks on board is not permitted. Oxygen can be provided only on certain flights for a fee. A request must be submitted at least 48 hours before departure. FAA approved concentrators can be used upon request however, because there is no power supply on board, you must carry a sufficient number of batteries for the flight duration. http://www.united.com/web/de-DE/content/travel/specialneeds/disabilities/oxygen.aspx
Virgin Atlantic	Taking along your own oxygen tanks on board is not permitted. Provision of oxygen may be available. A request must be submitted at least 72 hours before departure. There is the possibility to take own oxygen concentrators onboard. Please visit for more information: http://www.virgin-atlantic.com/gb/en/travel-information/special-assistance/onboard-oxygen.html

CHAPTER 13: SMALL MEDICAL DICTIONARY

6-Minute Walk Test – a simple way to test exercise tolerance, by measuring how far someone can walk in 6 minutes on the flat.

Air Compressor – is a machine that can suck air in and then eject it out under higher pressure.

Airways – the structures or passageways that direct air into and out of the alveoli.

Alveolar Proteinosis – a type of interstitial lung disease, characterised by defective or decreased surfactant production.

Alveolar – the terminal, grape-like protuberances of the bronchi, which represent the smallest structural unit of the lung.

Alveolitis - inflammation of the alveoli.

Alveolar Capillary Dysplasia (ACD) – a type of interstitial lung disease, in which there is abnormal development of the very small blood vessels surrounding the alveoli.

Anaesthesia – sleep induced by medication (sedation) with accompanying administration of analgesia (pain relief).

Antibodies – are special proteins produced by the body to help fight infection. They are specific for a particular pathogen and made post the initial contact with that pathogen (or vaccine).

Aspiration – the swallowing or inhalation of solids (e.g. food) or fluids into the lungs.

Arterial Hypertension – high blood pressure.

Arteries – the blood vessels that carry blood away from the heart.

Atherosclerosis – hardening and thickening of the arteries, with fatty deposition in the arterial walls.

Autoimmune Disease – diseases in which the person's immune system attacks their own cells

Azithromycin - an antibiotic in the macrolide family.
BCG – the *Bacullus Calmette- Guérin* vaccine is a live, attenuated (weakened) strain of the tuberculosis (TB) pathogen. Its use as a vaccine in children has not been recommended since 1998 because of potential complications.

BGA – *see* **Blood Gas Analysis**.

Biopsy – tissue removal from a living body.

Blood Gas Analysis – is a blood test to determine the level of oxygen and carbon dioxide in the blood.

Bone Marrow Transplant – is the transplantation of immature stem cells (precursor cells which can develop into multiple different cell lines) from the bone marrow. Stem cells from here are responsible for the formation of all the different types of blood cells.

Breath Sounds – the sound created in lungs by the turbulence of air flow during inhalation or exhalation, heard by means of a stethoscope. These normal sounds can be altered by diseases affecting the lungs resulting in distinct noises (e.g. wheezing).

Bronchi – large air conducting pipes/passageways of the airways.

Bronchial Asthma – a disease of the lungs in which the airways can spasm or become narrow in response to a known or unknown causes resulting in reduced airflow.

Bronchioles – the smallest conducting pipes/passageways of the airways.

Bronchiolitis – is inflammation of the bronchioles.

Bronchiolitis obliterans – is a multifactorial, inflammatory disease of the lungs resulting in scarring of the bronchioles.

Bronchitis – is inflammation of the bronchi.

Bronchoalveolar Lavage (BAL) – is a diagnostic procedure performed to examine cells, salts and microorganisms from the lungs; by flushing sterile saline solution (*salty water*) into the bronchi, sucking it out and then examining the resultant solution.

Bronchodilation – open the airways.

Bronchoscopy – is a procedure to look at the inside of the airways by passing a thin fibre optic cable (with a camera attached) directly down them via the mouth or nose.

Capillaries – are the smallest blood vessels in the body.

Cardiologist – is a doctor who specializes in diseases of the heart.

Carina – is the branching point of the trachea into the two main bronchi.
Cataract – is an opacification of the lens of the eye (i.e. it becomes more cloudy, less transparent).

Chemotherapy – a drug treatment that utilises chemicals that are particularly toxic to rapidly growing or dividing cells (notably cancer cells).

chILD – Children's Interstitial Lung Disease, or interstitial lung disease of childhood.

Chronic – lasting a long time or having a long duration.

Cilia – tiny, motile hair-like projections on the inner surface of the airways, involved with removing mucous and the smallest particles of dirt or dust out of the lungs.

Coeliac Disease (*Non-tropical Sprue*) – is a chronic disease of the digestive tract (small intestines) caused by intolerance to gluten, a protein that is commonly found in many cereals such as wheat, barley, oats, rye and spelt.

Computerized Tomography (CT) Scan – a special x-ray imaging technique using computer enhancement of serial x-ray slices (pictures). Formerly known as a *CAT scan*.

Cor pulmonale – is a disease state characterised by increased load or strain on the right-side of the heart (potentially from increased blood pressures in the pulmonary circulation). It may be acute or chronic.

Coronary Heart Disease – is heart disease caused by fatty deposition into the walls of the heart's blood vessels, narrowing them and hence restricting (or obstructing) blood flow through them.

Corticosteroids – a group of related messenger compounds (hormones), which are naturally produced in humans by the adrenal gland. They can also be synthetically produced and are often used as anti-inflammatories.

Cortisone – a member of the corticosteroid drug family (*see **Corticosteroids***).

CPAP - Continuous Positive Airway Pressure – a special type of ventilation.

Cyanosis – is blue discolouration of the lips or fingertips, and is a sign of low blood oxygen saturations.

Cystic Fibrosis (CF) – a genetic disorder which causes malfunction of various glands in the body (they notably produce abnormally thick, sticky mucous), leading to fibrotic scarring of the lungs and pancreas.

Diabetes mellitus – an illness characterised by chronically elevated blood sugar levels.

Dietician – a specialist in matters regarding proper nutrition.

Diffuse Panbronchiolitis – a rare and severe chronic lung disease with diffuse inflammation of the bronchioles, more common in Asian males.

Diffuse Parenchymal Lung Diseases (DPLD) – Another name for interstitial lung diseases.
Drain – a flexible, plastic tube used to remove excessive (or pathological) fluid or air from the body.

Dyspnoea – is the sensation of "shortness of breath", or difficulty in breathing.

Dystrophy – a degenerative disorder caused by inadequate or defective nutrition; e.g. **Failure to Thrive** or poor growth, and weight loss.

Echocardiography – an ultrasound investigation of the heart.

Emergency Medications (***Rescue Medications)*** – these are medications which should only be taken at home in case of a severe acute deterioration and are not normally taken regularly.

Emotional Lability - rapidly changing emotions (mood swings).

Endotracheal Tube – *or breathing tube*, is a plastic tube inserted directly down into the trachea to assist with breathing (mechanical ventilation), either by way of the nose or, more commonly, the mouth.

Erythrocytes – are the red blood cells which transport oxygen around the body.

Failure to Thrive – delayed growth or development in a child, often observed as not gaining or maintaining weight.

Finger Clubbing – is a clinical sign characterised by striking bulbous distention of the end segment of one or more fingers.

Flow Rate – the rate of movement of gas in litres per minute (L/min).

Flu – viral infection caused by the Influenza virus.

Food Diary – a diary, in which a person records each day all the meals, drinks and snacks they consume.

Fructose Intolerance or **Malabsorption** – is a disorder in which the body has difficulties digesting and absorbing fructose (the naturally occurring sugar in most fruit, some vegetables and honey).

Gas Exchange – is the enriching of oxygen from air (in the alveoli) and concomitant release of carbon dioxide in the opposite direction.

Gastric Tube – or *Feeding Tube*, is a flexible, plastic tube that is inserted into the stomach, so as to be able to give supplementary or total nutritional requirements (food and fluids) and some oral medications.

Gastritis – inflammation of the stomach lining (gastric mucosa).

Gastroenterologist – a doctor specializing in the treatment of diseases of the gastrointestinal tract.

Genetic – hereditary or inherited.
Glucocorticoid – *see* ***corticosteroids***.

Heart Rate – the number of heart beats per minute.

High Flow Nasal Cannulas – special nasal prongs which can deliver oxygen to the patient at rates up to 50L/min.

High Calorie Liquid Diet – supplementary drinks to boost dietary intake, which contain 1-1.5 kcal/mL.

Human Geneticist – is a doctor who specializes in the diagnosis of hereditary disorders.

Hydroxychloroquine – is a drug which was primarily used to treat malaria (broadly speaking, an antibiotic), but also has anti-inflammatory properties.

Hyperglycaemia – high blood sugar levels.

Hypertriglyceridemia – elevated blood levels of triglycerides (a type of fat).

Hypoxemia – insufficient blood oxygen levels.

Idiopathic – with no apparent cause.

Immunodeficiency – is a weakness of the immune system, which results in an increased susceptibility to infections and being more severely infected. In children this is more often an inherited weakness, rather than an acquired weakness.

Immunologist – is a doctor who specializes in diseases of the immune system.

Immunological – related to the immune system.

Immunosuppression – weakening of the immune system.

Immunosuppressive – a drug that weakens the immune system, insofar that transplanted cells (as in a transplanted lung) are no longer recognised as being foreign, and hence are not attacked by the immune system.

Inactivated Vaccine – *or killed vaccine*; is a vaccine that contains inactivated or killed viruses or bacteria, or constituent parts thereof. Exposure to these triggers the body to produce antibodies targeted against the source organism, without risk of the organism proliferating or reproducing.

Infectious – contagious, a source for spreading an infection.

Influenza – *see **Flu***.

In-patient – admitted as a patient in a hospital for treatment or investigations.

Intensive Care Unit – a ward or hospital department, that is equipped and staffed for the close supervision and management of critically unwell patients.

Interstitial – relating or pertaining to the narrow spaces between tissues or parts of an organ (e.g. the lung).

Interstitium – the tissue contained in the interstitial space (e.g. *connective tissue*)

Intravenous – directly into or through a vein into the bloodstream.

Intubation – is the procedure of inserting a flexible, plastic tube through the mouth or nose directly into the trachea (wind pipe) for artificial ventilation (breathing).

Invasive Ventilation – is a form of mechanically assisted breathing for a patient that requires a tube to be inserted into the trachea.

IV – *see **Intravenous***.

Lactose Intolerance - is a disorder in which the body has difficulties digesting and absorbing lactose (the naturally occurring sugar in milk).

Larynx – the voice box.

Listing – is the date at which your child's condition deteriorated to such an extent that they were added to the waiting list for an organ transplant.

Liquid Oxygen Systems – an oxygen supply in which the oxygen is stored in its liquid form in cylinders/tanks.

Live Attenuated Vaccine – is a vaccine created from a live pathogen, which has been weakened so as to become incapable of causing disease in a healthy person, but is still able to reproduce.

Lobe – the largest components of each lung unit.

Lung Biopsy – *see **Biopsy***.

Lung Function Tests - *see **Pulmonary Function Tests***.

Lung Transplantation – is a major operation that involves the removal of a patient's diseased lungs and replacing them with a pair of healthy (donor) lungs.

Macrolides – a subgroup of antibiotics.

Main Bronchus – is the left or right bifurcation (branch) of the trachea (wind pipe).

Mechanical Ventilation – breathing is performed by a machine, delivered either via a tracheal cannula or endotracheal tube.

Metabolic Disorders – are various congenital diseases resulting in different defects of metabolism (the process of breakdown, transformation and/or utilisation of nutrients or chemicals in the body).

Methylprednisolone – *see **Prednisolone***.

Nasal Flaring – is a sign of respiratory distress in infants and young children, characterized by widening of the nostrils whilst breathing in.

Nasogastric Tube (NGT) – a form of gastric tube that is inserted via the nose down into the oesophagus (food pipe) and then advanced into the stomach.

Nebuliser – is a device that can deliver a medication as a fine spray of very small particles, enabling deep inhalation into the bases of the lungs.

Neonatologist – a paediatric doctor who specializes in the treatment of premature and new-born children.

Neuroendocrine Cell Hyperplasia of Infancy (**NEHI**) – is a subtype of interstitial lung disease which mainly occurs in younger children.

Neuromuscular Disease – are diseases which adversely affect both nerves and muscles.

Nissen Fundoplication – a surgical procedure for reducing the backflow of contents (including acid) from the stomach into the oesophagus (food pipe).

Non-invasive Ventilation (**NIV**) – form of mechanical breathing support which does not require direct introduction of a breathing tube into the trachea (wind pipe).

Occupational Therapist – an allied health professional that assists patients with restoring or maximising skills of daily living.

Orthopnoea – is laboured, difficult breathing, often associated with the use of accessory respiratory muscles. Difficulties breathing unless sitting upright.

Osteoporosis – a disease associated with reduced bone mass (and density), resulting in increased bone fragility.

Oxygen Concentrator – is a device which collects room air and filters out the oxygen from the other gases (normal oxygen concentration is 21% at sea level), then concentrating the oxygen to ~90% content before delivering it to the patient.

Oxygen Nasal Cannula – a soft, plastic tube connected to an oxygen supply, which is then looped over both ears and has dual nasal prongs to deliver oxygen directly into the nose.

Oxygen Saturations – the amount of oxygen in the blood, which can be measured by a pulse oximeter.

Paediatric – area of medicine dedicated to the care of babies and children.

Palliative Care – is medical care that is directed at symptom relief (e.g. pain, nausea and dyspnoea), rather than cure. Its aim is to maximise quality of life.

Partial Pressure – is the pressure for each individual component of a gas mixture (e.g. oxygen in air).

Pathologist – a doctor who specialises in examining tissue samples.

Peak Inspiratory Pressure (PIP) – is the highest level of pressure applied to the lungs during inhalation. Increasing it may enhance oxygen absorption.

Peak Flow Meter – is a small portable device which can be used at home to measure the fastest rate a person can blow air out of their lungs (litres per minute).

PEG Tube – *Percutaneous Endoscopic Enterostomy Tube*, is a special gastric feeding tube that is passed directly into the stomach through a small incision in the left upper quadrant of the abdomen (belly).

Peptic Ulcer (**Gastric Ulcer**) – is an often painful condition associated with erosions (sores) of the stomach lining (mucosa); frequently caused by gastric (stomach) acid, a bacterium called *Helicobacter pylori*, stress or medications (e.g. *NSAID's, steroids* and *aspirin*).

PFT – *see* ***Pulmonary Function Tests***.

Physiotherapist (***Physical Therapist***) – is an allied health professional that helps patients regain or improve their physical activity through exercises, movement and education.

Pneumococci – a common bacterial cause of infections in humans (especially children). They can cause meningitis, pneumonia, middle ear and sinus infections.

Pneumocytes – are specialised types of cells lining the alveoli walls. Type I facilitate gas transfer and Type II produce the protective lubricant, surfactant.

Positive End Expiratory Pressure (PEEP) – is the lower, fixed pressure at the end of expiration in the lungs. Increasing this with ventilatory support aims to prevent lung collapose during expiration and hence improve oxygenation.

Prednisolone – an artificially produced corticosteroid that is effective in reducing inflammation.

Primary Ciliary Dyskinesia – is a rare, genetic disorder characterised by dysfunctional cilia in the airways.

Prognosis – an "educated" prediction on the most likely course or outcome of a medical condition.

Prophylaxis – is a measure attempting to prevent a disease or an infection.

Proton Pump Inhibitor (**PPI**) – is a class of medication used to reduce the production of acid in the stomach, hence used to treat (or prevent) gastritis or stomach ulcers.

Psychologist – is an allied health professional who specializes in studying and treating behavioural and emotional disorders in humans.

Pulmonary – relating to the lungs.

Pulmonary Arterial Hypertension – a condition characterized by high blood pressure in the pulmonary circulation.

Pulmonary Fibrosis – is a disease of the lung, where there is progressive scarring of the lung's interstitium (tissue between the alveoli).

Pulmonary Function Tests – are a group of tests that measure how well your lungs work.

Pulmonary Hypoplasia – is incomplete development of lungs, with resultant functional impairment.

Pulmonologist (**Respiratory Physician**) – a doctor who specializes in the treatment of diseases of the lung.

Pulse Oximeter – is a device which uses light to non-invasively measure the proportion of oxygen (saturation) in blood.

Pulse Oximetry – is the measurement of the oxygen saturation in blood.

Pulse Steroid Therapy – is the administration (often intravenously) of high dose steroids (corticosteroids) in an intermittent manner and usually maximally only over the course of days.

Radiograph – a digital image of the internal structures of the body, made by passing x-rays through the body onto an absorptive plate.

Radiologist - is a doctor who specializes in the implementation and evaluation of medical imaging techniques (e.g. x-ray, CT, nuclear medicine, magnetic resonance and ultrasound).

Reflux – is a medical condition involving the backward (retrograde) flow of the stomach's contents (especially acid) up into the oesophagus (food pipe). This is usually due to poor closure of the sphincter between the two organs.

Regular Medications – medications that are required to be taken regularly (e.g. every day).

Rehabilitation – is a treatment designed to help with the process of recovery after illness, injury or disease, and return to a functional level appropriate for everyday life.

Rehabilitation Physician – is a doctor who specialises in the prevention, alleviation and recovery from chronic diseases, conditions or illnesses.

Respiratory – is relating to the process of gas exchange in living organisms.

Respiratory Failure – is a condition in which the body can no longer compensate for the limitations of poor blood oxygen levels, often resulting in the signs of cyanosis, respiratory distress, and reduced or altered levels of consciousness.

Respiratory Rate – is the number of breaths per minute.

Respiratory Syncytial Virus (**RSV**) – is a common viral cause of upper respiratory tract infections, such as colds, acute bronchitis, and middle ear infections.

Restrictive Ventilatory Defect – is a lung disorder which is characterised by having reduced lung elasticity, with resultant restricted lung expansion or retraction.

Retractions (or **Recessions**) – are a sign of respiratory distress in children, where there is "*indrawing*" (the sucking in) of the skin between the ribs (***intercostal recession***), above the collar bones (***supraclavicular retraction***) or between the collar bones (***jugular retraction***) with breathing in (*inhalation*).

Rheumatologist – a doctor who specializes in treating inflammatory disorders of joints, muscles, bones and connective tissue.

Right Lung *and* **Left Lung** – are the base or composite units of the lungs.

RSV – *see* ***Respiratory Syncytial Virus***.

Saturations – are the proportion or relative concentration of something dissolved into another medium, usually given as a percentage. For lung disease this term is used for the amount of oxygen in the blood.

Sedation – the act of giving a medication to produce a state of calm or sleep in the recipient.

Skin Prick Test – is used to test for a reaction to allergens.

Social Worker – an allied health professional with special training and knowledge of available social support services (e.g. they can provide advice for gaining financial or care assistance).

Specialist – *in the context of medicine*; is a doctor with more extensive knowledge and training in a particular field or area of medicine.

Speech Therapist – an allied health professional that assists patients with assessing and treating speech and swallowing disorders.

Steroids – a group of medications that can reduce inflammation, treat allergic reactions and treat cancer. *See* ***Corticosteroids***.

Surfactant – *Surface Active Agent*; is a thin film of protein-rich fluid covering the surface of alveoli, which prevents the alveoli from collapsing and sticking together during exhalation.

Surfactant Metabolic Diseases – a group of diseases characterised by defective production or breakdown of surfactant.

Survival –is the percentage of patients alive after a certain amount of time (*after diagnosis* or *post lung transplant*).

Sweat Test – a test for cystic fibrosis, checking for a higher concentration of salt (sodium chloride) in the sweat.

Systemic Disease – are diseases that affect more than one organ or part of the body

Tachypnoea – rapid breathing, increased respiratory rate.

Therapeutic – related to treating or curing a disease.

Thorax - Chest

Trachea – wind pipe.

Tracheostomy Tube – a special plastic or metal tube inserted surgically into the trachea, through an incision in the skin just below the voice box.

Tracheal Stenosis – narrowing of the wind pipe.

Tracheomalacia – is a condition in which the cartilage rings of the trachea are too soft, resulting in partial or complete collapse of the wind pipe.

Tracheotomy – is the surgical creation of an opening directly into the trachea through the neck (beneath the voice box), so as to relieve difficulties in breathing (including the provision of invasive ventilation)

Transplant Team – is a team of physicians, nurses, psychologists, therapists and social workers, all of whom are responsible for the pre-operative preparation, operation and post-operative management of a transplant.

Vaccination – a medication that is intended to protect against infection and is usually given as an injection.

Veins – blood vessels that carry blood towards the heart.

Video Assisted Thoracoscopic Surgery (VATS) – is a surgical procedure in which a camera is inserted through a small incision into the chest, enabling doctors to look at the surface of the lung and also to potentially take a lung biopsy.

CHAPTER 14: APPENDIX

Checklist for the First Visit to the Specialist Centre:

□ Letter of Referral
□ Health-Insurance Card
□ Vaccination Record/"*Personal Child Health Record"*
□ All previous medical or discharge letters
□ Previous Pulmonary Function Test results
□ Results from prior laboratory investigations (including blood tests)
□ Images from previous Chest X-rays (on CD)
□ Images from any previous CT-thorax scans (on CD)
□ Report or findings of any prior Echocardiograms
□ Results of any prior Sweat Test
□ Names and contact details of previously involved treating doctors
□ Current treatment plan (including a list and doses of current medications)
□ All other required documents as per appointment letter/telephone booking
□ A prepared (written) list of specific questions you have for the doctor

Emergency Action Plan (*Emergency note for Doctors*)

1. Name:

2. Age and Date of Birth:

3. Recent Weight:

4. Diagnosis:

5. Requiring Oxygen: □ yes □ no
If so, at what flow rate? L/min

6. Requiring Ventilation: □ yes □ no
If so, what are the settings?

- PIP:
- PEEP:
- FiO2:
- Device:

7. Special Instructions for an emergency:

8. Emergency (rescue) Medication and dosage:

9. Name and Telephone Number for the parents:

10. Other important contacts/phone numbers/addresses (e.g. attending physicians, current treating centre):