Diagnosing Childhood Interstitial Lung Disease

Lung Foundation Australia

Diagnosing childhood Interstitial Lung Disease (chILD) is often difficult because these diseases are rare and have symptoms that are similar to many other lung diseases. Many doctors will not be familiar with some of the diseases or may think that the symptoms are caused by a more common disease, like asthma. As a result, the diagnosis is often delayed. Sometimes months or even years pass between the start of symptoms and the final diagnosis. Given the rarity of these conditions, it is also possible that the specific cause, or diagnosis will remain unknown.

Paediatric respiratory specialists are best placed to diagnose chILD

Early diagnosis and treatment are important for all types of chILD. Due to the difficulties in diagnosing these groups of rare lung diseases and the importance of obtaining the most accurate diagnostic information, your child should be seen by a paediatric respiratory specialist with expertise in the diagnosis and management of rare lung disease.

Parents have a key role in supporting accurate diagnoses

Parents and carers can support accurate diagnoses by maintaining detailed records of their child's medical history and symptoms. Every individual characteristic of your child's disease can help to identify the specific type of chILD and determine the best treatment. Taking a video of your child's breathing and symptoms to show doctors is often very helpful.

Steps in diagnosis

Referral to a paediatric respiratory specialist

It is important that your child is referred to a paediatric respiratory specialist.There is not one specific test used for diagnosis. The paediatric respiratory specialist will conduct a number of diagnostic tests to exclude other conditions with similar symptoms and to help determine your child's diagnosis.

Currently there are no best practice guidelines for the diagnosis, treatment and management of chILD in Australia. Increasingly, a team of specialists work together to establish the most likely diagnosis under a network called chILDRANZ (Children with Interstitial Lung Disease Research -Australia and New Zealand). The network is made up of multidisciplinary health professionals from all over Australia with a clinical and research interest in chILD. It meets monthly to discuss cases of children with suspected chILD. Ask your doctor to connect in with this network if they are not already involved.

Detailed examination

The paediatric respiratory specialist will spend time with you and your child undertaking a detailed examination. This will include taking a detailed medical history of your child and family, conducting a thorough physical examination including measurement of body weight and height, and measuring your child's breathing rate. A detailed examination is an essential component for successful diagnosis. Doctors will ask questions such as:

- What symptoms are present?
- When did the symptoms start?
- What tests/investigations have been performed?
- What treatments have been given so far and how did your child respond?
- Has your child been treated in hospital?
- Is there a family history?

Records to take to your first visit at a specialist centre:

- Your child's prior medical history – where possible, presented in chronological order of each of the events and tests undertaken. This may be presented as a list, or diary format.

- Copies of all previous medical letters and results of previous lung function tests.
- Copies of all prior X-rays and CT-scans. You can ask for these to be transferred electronically to the clinic. Some may be stored and available via the local radiology storage system so check with the paediatric respiratory specialist before the appointment.
- Some of your child's signs or symptoms may only occur intermittently (and never with a doctor present) and may be difficult to describe. If you are able to video your child when these signs or symptoms are happening, bring these recordings with you to the visit.

You can use My Health Record to keep track of your child's health information online. If your child doesn't already have a My Health Record, you can register for one at any time.

Excluding other conditions with similar symptoms

Before being able to diagnose chILD, other conditions may need to be excluded. Some conditions with a similar presentation to chILD that may need to be excluded:

- Cystic fibrosis
- Asthma
- Congenital malformation of the airways
- Congenital malformation of the lung
- Congenital heart disease
- Chronic infections of the airways
- Aspiration and gastro-oesophageal reflux
- Immune deficiencies, auto immune disorders and other disorders of immune function
- Rheumatological disorders
- Functional disorders of the cilia (e.g. primary ciliary dyskinesia)
- Idiopathic pulmonary arterial hypertension

Common tests

There are a number of tests or investigations that are performed in all children suspected of having chILD. These tests or investigations can also help to diagnose or rule out other conditions with similar symptoms.

Common tests and investigations in the diagnostic process include:

Image techniques

Chest X-ray

A simple X-ray can provide the doctor with basic information regarding the organs within the chest (thorax) including heart, lung and blood vessels. Particular 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 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.

Computed Tomography (CT scan)

The CT-scan is the most important radiological imaging technique for suspected chILD. It will provide more detailed information than a plain X-ray. Although it is unusual for a CT-scan to establish the precise diagnosis, the images can confirm the presence of chILD and help plan the best site to obtain tissue samples if a lung biopsy is to be done. A disadvantage is the higher radiation dose than a chest X-ray, though new modern-day fast scanners have much lower radiation doses.

If your infant or toddler is having a CT scan, the doctor may recommend a sedative, or even a general anaesthetic, to keep your child comfortable and still. Movement blurs the images and may lead to inaccurate results. Ask your doctor how to prepare for the test with your child. At present there is no worldwide standardised protocol for performing CT scans in chILD.

Echocardiogram

An echocardiogram (or 'echo') is an ultrasound of the heart. An echo uses sound waves to produce images of your heart. Your doctor can use the images from an echocardiogram to identify heart disease.

Lung Function Tests

Lung function tests give valuable insight into the type and functional impact of lung disease as they measure how well the lungs are working. Lung function tests help to confirm the presence of chILD. They are not painful but can be quite tiring as they may require repeated breaths in and out at maximum force and depth – they are the most important test in many cases for monitoring progression of disease and response to treatment.

Many lung function tests are usually only performed in children from the age of five, as the child needs to be able to understand and follow instructions. There are not many places in Australia who can perform infant lung function tests in children under one as they are mainly done for research purposes currently.

Tests for gastro-oesophageal reflux

Barium Swallow

Barium Swallow tests allows the examination of the digestive tract using a white powder (mixed with water) which appears on an X-ray. A radiologist can evaluate a

wide range of problems that may be the cause of your child's symptoms including: areas of reflux, narrowing or ulceration to the digestive tract.

Modified Barium Swallow (MBS)

Modified Barium Swallow (MBS) is a fluoroscopic procedure designed to determine whether food or liquid is entering your child's lungs, also known as aspiration. MBS is an analysis of swallowing through three phases: oral (mouth), pharyngeal (throat) and upper oesophagus. It permits the treating team to observe the coordination of anatomical structures in the mouth and throat, as they actively function when chewing, drinking and swallowing. It also identifies the reason for aspiration. It is usually done by a speech pathologist who will assess your child's swallowing.

Oesophageal pH monitoring

Oesophageal pH (acidity) monitoring tests can detect whether stomach acid is refluxing into the oesophagus, causing the pH to drop. Both 24-hour and wireless oesophageal pH monitoring are options. During oesophageal pH monitoring tests, you will be asked to record your child's food intake, sleep times and any symptoms your child experiences. The test can help determine whether the acid level in the oesophagus is linked to your child's symptoms.

Impedance reflux testing

Impedance testing can detect the reflux of non-acidic or less acidic stomach contents into the oesophagus. Impedance reflux testing is sometimes combined with 24-hour pH monitoring. It involves the same type of procedure as a 24-hour pH test. It measures liquid movement from the stomach into the oesophagus.

Endoscopy

Endoscopy is another procedure for investigating individuals who may have chronic gastro-oesophageal reflux disease (GORD). By placing a small tube with a light and tiny video camera on the end (endoscope) into the oesophagus, inflammation or irritation of the tissue lining may be seen. It is carried out under general anaesthesia or sedation.

Blood tests

Blood tests

During initial investigations, your child will have many blood tests done and many of them may be repeated. Beyond genetic tests, blood tests don't usually confirm or exclude a diagnosis of chILD. However, results of the blood tests can offer important indicators to confirm or exclude certain diseases.

Blood gas test

This blood gas analysis test is most often performed by taking a sample of blood from the tip of the finger. The purpose is to measure the levels 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 condition.

Measurement of oxygen saturations

Pulse oximetry

Pulse oximetry is a slightly less precise way of measuring arterial blood oxygen levels but is not painful or invasive. It is performed with a superficial skin probe (pulse oximeter). It can be attached to either a finger or ear lobe (with a Band-Aid) and then connected to a monitor via a cable.

Pulse oximetry is easy to use to monitor oxygen saturation levels. It also shows the heart rate. Some monitors can store these measurements over a period time (e.g. 24 to 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. Pulse oximetry can also be used overnight to monitor and record oxygen levels as a child sleeps. Some children may require a formal sleep study.

Sleep study

A sleep study can help to diagnose sleep problems. It is an overnight test that can record a variety of functions whilst a child sleeps such as heart rate, breathing patterns, oxygen and carbon dioxide levels, snoring, body movements and sleep positions. The sleep study is usually performed by a sleep specialist at a dedicated sleep clinic.

Exercise capacity

6-minute walk test (6 MWT)

The 6 MWT is an exercise test and provides information regarding functional capacity, response to therapy across a broad range of chronic cardiopulmonary conditions. Main strengths of the 6 MWT stem from its simplicity in concept and performance, low cost, ease of standardisation, and acceptance by patients. It is a basic investigation for children over the age of five.

Cardiopulmonary Exercise Testing (CPET)

Cardiopulmonary Exercise Testing (CPET) is a non-invasive method used to assess the performance of the heart and lungs at rest and during exercise. The full cardiopulmonary system is assessed during a CPET by measuring the amount of oxygen your body is using, the amount of carbon dioxide it is producing, your breathing pattern, and electrocardiogram (ECG) while riding a stationary bicycle or running on a treadmill. This test may be performed in children over eight years of age.

Sweat test

A sweat test is used to diagnose Cystic Fibrosis (CF). People with CF have a high level of chloride in their sweat

which is identified during a sweat test. CF is another medical condition with similar symptoms to chILD which may need to be excluded when making a diagnosis.

Genetic tests

Only some of the chILD diseases have known genetic causes. Blood tests and other tissue samples are used to look for known genetic disorders or to discover newly associated genes. These tests can take weeks to months to come back. They give a clear, precise diagnosis without the need for lung biopsy (when positive). However, only a few types of chILD can be diagnosed this way including:

- 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)

You should see a genetic counsellor before these tests are undertaken. The results can also be discussed with a counsellor. If your child has a genetic cause for chILD it may impact on your extended family. It is useful to discuss with a genetic counsellor how to approach other family members.

Bronchoscopy

Bronchoscopy is a procedure in which doctors are able to look directly inside the lung by inserting a thin cable with a light and a small camera into the airways.

A bronchoscopy allows doctors to examine the airways for any malformation, being too floppy/soft or narrow, excessive secretions or signs of inflammation in the airway. Saline may be introduced into the airways and removed by suction. It is then examined for markers of infection, inflammation and other cells.

A bronchoscopy is always done under anaesthetic in children and young people. It is a safe and well-tolerated procedure.

Lung biopsy

Currently, this is the "gold standard" test used to diagnose and type many forms of chILD. A lung biopsy involves removing small pieces of abnormal lung tissue to examine under a microscope.

This procedure is done in select cases, where other investigational steps have not revealed a diagnosis. It requires a general anaesthetic and admission to hospital. It is usually done using a keyhole technique with small cuts on the side of the chest to insert a telescope and specialised instruments to take several pieces of lung, each about the size of a cherry. Generally, lung biopsies are only done when the test result will make a difference in treatment choices or will provide important information about diagnosis and outlook.

An experienced pathologist can learn a lot about the health of the lung tissue and identify many of the chILD diseases from the lung biopsy. However, a biopsy does not always yield a diagnosis. Biopsies should be performed at specialist centres experienced in caring for children with chILD whenever possible.

FURTHER INFORMATION AND SUPPORT

Contact Lung Foundation Australia for more information, to access our support services and join our mailing list for regular updates and latest news.

Lung Foundation Australia Services

- Information and Support Team
- Lung disease information resources
- Education webinars
- Lung Cancer and Respiratory Support Nurses
- Support groups
- Peer-to-peer connections
- Referral to pulmonary rehabilitation and Lungs in Action exercise programs
- E-newsletter

External Links

- Australian Genomics
  www.australiangenomics.org.au
- Consumer Information
  www.genomicsinfo.org.au
- US Child Foundation
  www.child-foundation.org

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