

# An Introduction to Cystic Fibrosis for parents and families



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## Introduction

This booklet has been put together by the Cystic Fibrosis team at Monash Medical Centre to assist you during the time of diagnosis.

By now you would have met with members of the team (doctor, coordinator, social worker, physiotherapist, and dietitian). The team would have begun discussing CF and its management with your family. At times, especially during this first week, this information can feel confusing. Families often say they feel tired during these first few days and often how difficult it is to remember what has been discussed with the team during the time of diagnosis.

The team is very aware of the need for families to have the opportunity for frequent and regular discussion with all team members and we encourage you to do this with us. The team acknowledges that you know your child best and we are committed to working with you in managing your child's CF.

The team is also aware that everyone adjusts to new information at their own pace and that information may need to be discussed many times before a family feels confident and comfortable. Many families find it helpful to write down any questions they have for the team. Feel free to do this as often as you need to. As you know your family and your child best, we will be guided by you in how often you need to discuss CF information.

## What is Cystic Fibrosis?

Cystic Fibrosis (CF) is an inherited disease that affects the fluids (or secretions) of the sweat glands, lungs, pancreas and bowel. The liver and the male reproductive system are also affected. People with CF have a change in the amount and composition of secretions in these organs, that leads to a blockage within the affected organ. The consequences of this blockage depend upon the organ involved, as shown in Table 1.

<b>Organ</b>	<b>Result of Blockage</b>
Lung	Infection, inflammation and lung damage
Pancreas gland	Unable to digest food, diabetes
Bowel	Constipation, bowel obstruction
Sweat glands	Increased salt levels in sweat
Liver	Elevated liver enzymes, liver damage
Male reproductive system	Infertility

It is also important to understand that many organs of the body are not affected by CF at all. These include the heart and the brain. Therefore CF does not cause brain damage and all children with CF will attend normal school and their level of academic achievement should be no different to their non-CF peers.

Although there is currently no cure for CF, advances in medical treatment have dramatically increased survival, and now we expect all newborn babies with CF to live full and productive lives well into adulthood. We expect that the current average survival of 37 years to continue to improve as newer treatments become available.

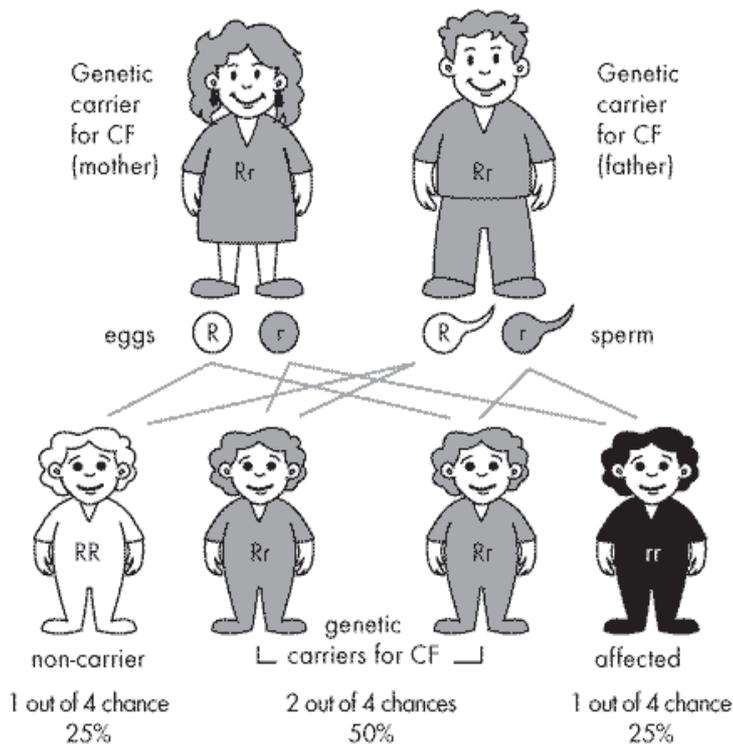
Recently, new drugs have been developed that improve the function of the CF protein [CFTR – see below]. These new drugs [Kalydeco and Orkambi] are not yet widely available in Australia due to cost, however in time we expect this to change, and many adults and children with CF will benefit,

Eventually, gene therapy for CF lung disease will be available. Until that time, the Monash CF team will work with you to ensure that your child maintains optimal health at all times.

## Genetics of Cystic Fibrosis

CF is an inherited genetic disease. This means that CF is caused by a change or mutation in the CF gene. The gene involved in CF controls the production of a protein called CFTR or **C**ystic **F**ibrosis **T**ransmembrane **C**onductance **R**egulator. This protein controls the movement of salt (sodium chloride) in and out of cells in the body. When a mutation occurs in the CF gene, the function of the CFTR is abnormal, which results in an abnormality in the movement of salt in or out of cells. This abnormality then leads to the problems noted in Table 1. There are more than 1500 different mutations in the CF gene, but the most common in Australia is called  $\Delta F_{508}$ .

Like other genes in the body, the CF gene exists as a pair. Any individual inherits one gene from each parent. The inheritance pattern in CF is known as autosomal recessive. For CF to occur in any newborn child, each parent must be a "carrier" of a CF mutation. This means that in each parent, one CF gene is normal and the other carries a mutation. When each parent passes on their abnormal CF gene, the child will have 2 copies of the abnormal gene, and will be affected with CF. If only 1 parent passes on an abnormal CF gene the child will be a carrier and completely healthy. If neither parent passes on their abnormal CF gene, the child will have completely normal CFTR function and will not be a carrier (Figure 1).



*Figure 1. Cystic Fibrosis is an autosomal recessive condition where both parents are unaffected carriers for CF. Both copies of the abnormal CF gene have to be present for the individual to be affected with CF*

## Diagnosis of Cystic Fibrosis

### 1. Newborn Screening

In Australia, the most common way in which CF is diagnosed is via the Newborn Screening Program. This involves the analysis of the heel-prick blood spot card (also known as the Guthrie card), which is obtained from every 4 day-old baby born in Victoria (a similar system operates in all other states). The blood spot is analysed for levels of trypsin, which is one of the digestive enzymes produced by the pancreas. In those

without CF, the pancreatic cells produce trypsin, which then flows along small tubes (or ducts) within the pancreas, until the main pancreatic duct which connects to the gut. In this way, the digestive enzymes produced by the pancreas mix with food and allow digestion to occur. However, in babies with CF, the pancreatic secretions are abnormal, which causes blockage of the pancreatic ducts. Thus, trypsin produced by the pancreas is prevented from entering the gut, and instead spills over into the blood stream, resulting in greatly elevated blood levels of trypsin, which is then detected in the heel prick blood spot.

Those babies with elevated trypsin levels in the Guthrie card blood spot then undergo CF gene mutation testing from the same blood spot. If 2 CF mutations are identified, the baby is affected with CF and the treating doctor is notified by Genetics Victoria [GV]. The baby is then referred to the CF service of either Monash Children's Hospital or the Royal Children's Hospital (depending on postcode). If only one mutation is identified, the baby may be a carrier or may have a second mutation that is not detected by the screening program. Remember there are over 1500 other possible mutations, so rather than test for all of these, the GV laboratory tests for the 12 next most common mutations. If none of these are found, the parents are contacted by GV and advised that an abnormality has been detected on the newborn screen, and that a sweat test is necessary to either confirm or rule out the diagnosis of CF. The sweat test is performed by the biochemistry laboratory at Monash Medical Centre. The diagnosis of CF is made if the sweat chloride level is greater than 60 millimoles per litre.

## 2. Meconium Ileus

Between 15 and 20% of newborn babies with CF develop a bowel obstruction at birth known as meconium ileus. In this condition, the baby fails to pass meconium in the first 24 hours after birth and develops a swollen abdomen and vomiting. An abdominal X-ray shows a small bowel obstruction. Sometimes an enema can clear a mild obstruction, but usually an operation is required. An ileostomy or colostomy bag may be required for a number of weeks after the operation, depending on the level and severity of the obstruction. Eventually the ileostomy is closed and the bowel function returns to normal.

## 3. Clinical Features

The current newborn screening program for CF is very effective, with over 95% of affected infants correctly identified. However, the program is not perfect (no screening program is ever perfect!) and therefore, on average the program will miss 1 or 2 affected CF infant per year in Victoria. These children develop symptoms of either CF lung disease (a chronic recurrent wet cough), or failure to thrive because of CF pancreatic disease (diarrhoea, abdominal bloating and poor weight gain). The presence of these symptoms usually leads to the suspicion of CF, which is confirmed by sweat testing.

## Management of newly diagnosed infants with Cystic Fibrosis

### The Multidisciplinary CF team

Research and experience around the world has shown that the best health outcomes for children, adolescents and adults with CF are best if care is delivered by a multidisciplinary CF team. At Monash CF, the team members include doctors, physiotherapists, dietitians, social worker, nurse, psychologist and a CF clinic coordinator. The family's attendance at 'clinic' or outpatient appointment is an opportunity to discuss your child's care with all members of the CF team.

Once the diagnosis has been confirmed, an initial meeting will be made for you to meet with one of the CF team doctors. At this meeting, the diagnosis is explained, your first questions answered and parental and family education arranged. You will meet with the CF physiotherapist, who will explain and demonstrate techniques of chest physiotherapy that you will do to keep your baby's chest clear of secretions. The CF

dietitian will teach you how to give pancreatic enzymes before feeds. Regular meetings with your child's CF doctor during this time will ensure that any questions you have are answered promptly.

Following initial education, most newly diagnosed CF infants will be reviewed in the outpatient clinic every 1-2 months to check on weight gain and to monitor lung health. The Monash CF team strongly encourages you to become involved in all aspects of your child's CF care. Team members are contactable by phone and email and welcome any questions you have.

## **How does CF affect the lungs?**

Babies with CF have normal lungs at birth. The abnormal CF gene causes a change in the volume of the very thin layer of fluid secretions, which normally lines the airways of the lung (bronchi). This change causes the fluid to become thick and sticky, leading to blockage of the small airways. Unless the airway secretions are removed, infection of the airway eventually occurs, leading to bronchitis. Recurrent or persistent bacterial bronchitis leads to lung damage. Untreated CF lung disease eventually causes death from respiratory failure. The aim of treatment of CF lung disease is to ensure that the airways of the lung are kept clear of secretions, and episodes of bacterial infection are aggressively treated with antibiotics.

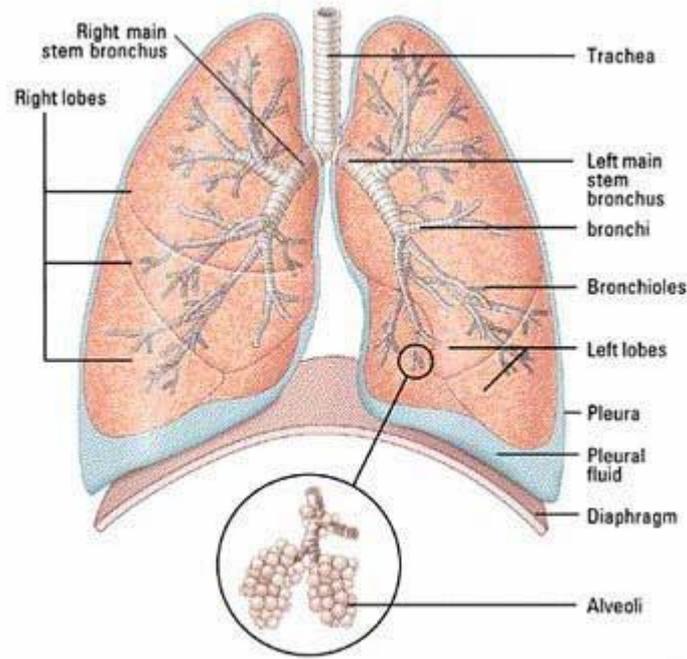
## **Chest Physiotherapy**

Regular daily chest physiotherapy is an essential part of the management of CF lung disease. You will be taught how to perform chest physiotherapy for your baby by the CF team physiotherapist. We expect that all babies with CF will have chest physiotherapy performed once or twice a day when they are well and more frequently if they become unwell with a chest infection. As your child grows older and can understand how to co-operate, they can be taught to perform various chest physiotherapy techniques for themselves. We also strongly advocate that all children with CF should maintain a high level of physical activity, and so we encourage full participation in any and all sports that are available or of interest.

Chest Physiotherapy in people with CF is a generic term used to describe techniques which clear sputum from the lungs (airway clearance techniques), exercise and activity as well as encompassing the use of inhaled medications.

Chest physiotherapy treatment is both preventative and for the management of respiratory symptoms, and needs to be performed at least daily. It will include many different techniques or activities dependent upon your child's health, age and stage of development. Early after your child's diagnosis you will be taught techniques to continue this daily treatment at home.

## **The Lungs**



### Why does my baby need chest physiotherapy?

We all have mucous (sputum) in our lungs. In people with CF this mucous is thicker and stickier than normal making it harder to clear. If mucous becomes trapped in the lungs it can lead to infection and thus inflammation. This in turn produces more mucous, which is thicker and stickier than normal, leading to a “vicious cycle” within the airways (*see figure*). Regular chest physiotherapy is intended to keep the thick and sticky mucous moving, and help prevent the airways becoming blocked (“obstructed”).

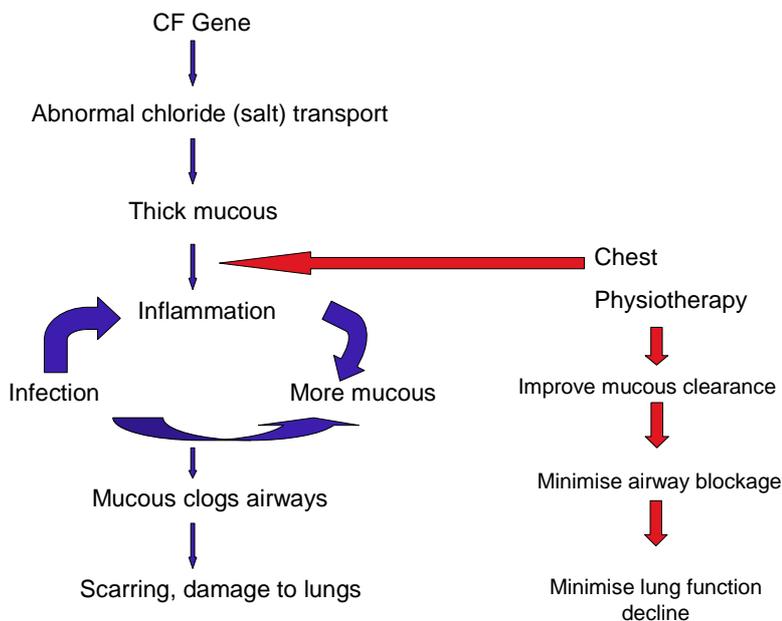


Figure: Why chest physiotherapy is important in the treatment of Cystic Fibrosis Lung Disease

### What evidence is there for the benefits of regular chest physiotherapy?

Physiotherapy is commenced in the period following diagnosis for a number of reasons which include:

- Evidence suggests that even in asymptomatic infants inflammation, infection and structural changes exist in the airways<sup>1,2</sup>. Treatment of early changes may preserve lung function and optimise long-term outcomes<sup>3,4</sup>.
- *Airway clearance techniques* aim to improve mucociliary clearance – that is the movement of sputum (mucous) out of the lungs- so that the development of chronic lung disease may be delayed<sup>5,3</sup>
- Establishing an early routine may assist with adherence and teaches effective airway clearance techniques for when the infant is acutely unwell
- Early, regular treatment can help parents/carers to recognise signs and symptoms of an acute illness, so that necessary treatment can be started quickly.

### What do we do for chest physiotherapy?

It is aimed that airway clearance for the newborn should fit in with the family's routine. General consensus amongst Australian physiotherapists working with children with CF suggests treatment of an asymptomatic infant should consist of 4-5 modified postural drainage positions performed 1-2x/day. In each position percussion should be performed for 3-5 minutes.

- **Modified Postural Drainage:** Modified postural drainage refers to the positions we get you to place your baby in whilst performing percussion (see below). These different positions allow gravity to help drain mucous in the lungs toward the large airways. The 5 most commonly used positions are:
  - Baby lying on their right side
  - Baby lying on their left side
  - Baby lying on their back
  - Baby lying on their tummy
  - Baby positioned in upright on resting on your chest/shoulder
- **Percussion:** Percussion is *firm* patting of the chest using the fingers – or a cupped hand in older babies – over a folded towel, other cloth or adequate clothing. When we percuss the external, mechanical pressure we provide to the chest wall helps mucous to move from the small to larger airways.
- **Vibrations:** During “out” breaths (exhalation) vibrations (or “vibes”) can be used. These are gentle, but firm, “squeeze and shake” movements generated with your whole arms through your hands. You may not be taught vibrations when your child is first diagnosed, but rather later, nearer to 1 year of age.
- **Cough:** Infants will cough spontaneously if they need to, and will generally swallow any mucous (sputum) which they cough. Occasionally, if a child is quite “chesty” and has increased cough they may vomit some of this mucous. Infants with CF do not need to be provoked to cough. As children age they can be taught to cough on command.

It is important to note that other techniques may be used for appropriate patients.

### When should we do chest physiotherapy?

Chest physiotherapy is recommended to be completed prior to feeds, or at least 1 hour after feeds. The child should be awake when the treatment is commenced, and should be performed at a time that best fits in with family routine.

### How often should we do chest physiotherapy?

As a general rule, a well infant with no symptoms will need 1-2 sessions of chest physiotherapy a day lasting 15-25 minutes. This may be increased if your child is unwell, or while you are learning the techniques. A child who is sick in hospital may have more frequent sessions.

### Exercise

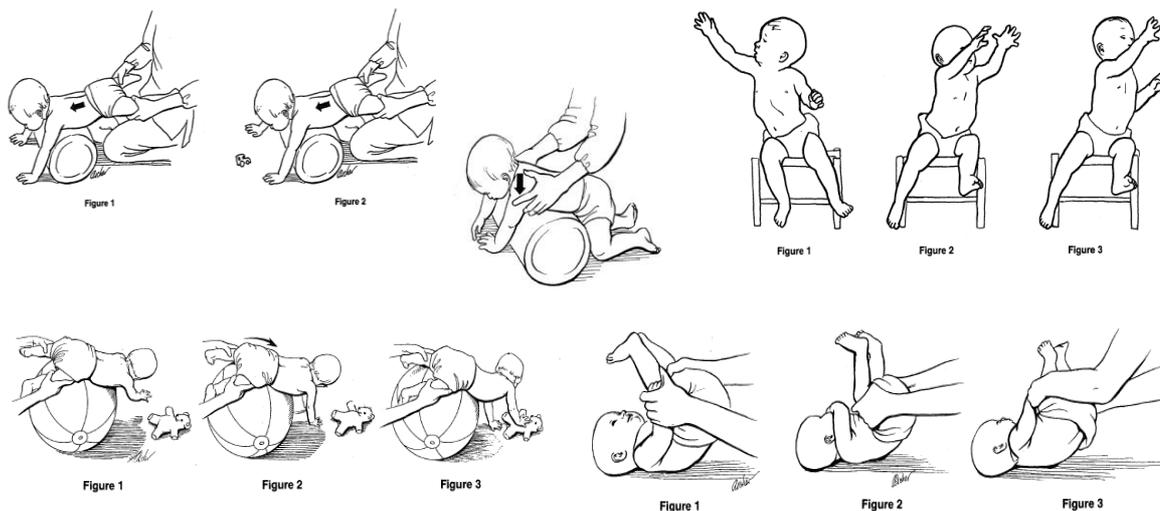
Whilst it is generally accepted that airway clearance techniques provide the basis for assisting in sputum clearance and maintaining lung health, regular exercise is vitally important.

Exercise should be considered and encouraged as part of overall physiotherapy management in CF. From time of diagnosis, irrespective of age, exercise and physical activity should be incorporated into the daily routine. As a young child, family and siblings should be encouraged to also be involved in activity and exercise to assist in normalising the activity and making it 'fun'. Exercise is important not only for maintaining aerobic fitness, but also creating strong bones and good posture.

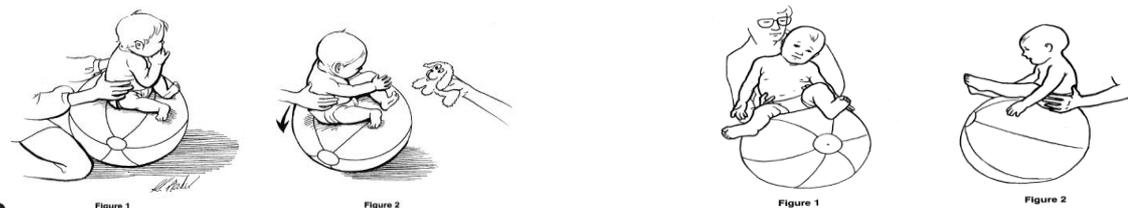
Whilst not exercise, the playing of a musical (wind) instrument, or singing, are other activities that can help to maintain the health and function of your child's lungs.

To follow is a collection of suggestions of activities and postures aimed to encourage developmental play, thoracic range of motion, increased lung volumes through upper limb activities and FUN!!

- **Rolling, Crawling and Reaching:**



- **Ball games, Bouncing and Jumping:**



### Trouble-shooting:

Some babies will fall asleep during physiotherapy, whilst others will seem to be uncomfortable, squirming and crying. A baby may even change from being “easy” and settled with physiotherapy to crying and annoyed. This often coincides with progression of your baby’s development as they become more active and alert. Once they start to explore by crawling and walking, physiotherapy time can become quite challenging.

It can be worth spending a few minutes doing settling activities (rocking, singing, cuddling) before starting chest physiotherapy treatment. It is natural for an adult to stop patting a baby’s chest if s/he starts to cry. Try to prevent this trap by watching and anticipating when they’re about to cry, possibly changing baby’s position, but not stopping completely.

Remember, as the parent, you need to maintain control of the session. Some days will be easy and others difficult, but a variety of strategies can be employed to keep things positive. Your patience, creativity, flexibility and sense of humour can make chest physiotherapy sessions effective, acceptable and enjoyable to your child.

### **Where to next?**

As your child gets older your Physiotherapist will introduce you both to more active forms of treatment. These may include activities like bubble blowing, bouncing on a trampoline and tickle games. By the time your child heads off to school we would hope they are completing a more independent form of chest physiotherapy with your supervision. As your child gets older regularly ask your clinic physiotherapist if there are any changes you should anticipate over the next few months with respect to your child’s chest physiotherapy.

### **A final word:**

Physiotherapy can be a special time, when your child can have your undivided attention. You need to be confident in what you’re doing, and remember that cuddles, smiles and laughter are as much a part of the therapy session as percussion and positioning.

### **Antibiotics**

Antibiotics are used to manage episodes of chest infection. The most common symptom of a chest infection in CF is a persistent wet rattly cough. This usually starts with a runny nose and mild fever. Once the cough appears, we would advise treatment with oral antibiotic syrup, and increased chest physiotherapy. Usually this regime is undertaken for 7-14 days, until the cough disappears. If the cough gets worse despite treatment, your CF doctor may recommend either a different oral antibiotic, or possibly admission to Monash Children’s at Monash Medical Centre for intravenous antibiotics and intensive chest physiotherapy (often called a “tune-up”). During this admission, your CF doctor may recommend an investigation called flexible bronchoscopy – this involves your child having a short general anaesthetic while a fibre-optic bronchoscope is passed into the airways of the lung. Saline is then injected through the bronchoscope, and sucked back into a specimen container. This lung washing is then sent to the hospital microbiology laboratory for culture. This identifies the exact bug causing your child’s infection, allowing your CF doctor to prescribe the ideal antibiotic combination to eradicate the infection. A hospital tune-up for CF usually lasts 7-14 days, depending upon symptom severity.

### **Bacteria Causing Lung Infection in Cystic Fibrosis**

Only a small number of different types of bacteria cause lung infection in people with CF. In babies and pre-schoolers, the two most common organisms are *Staphylococcus aureus* and *Haemophilus influenzae*.

Infection due to these organisms can be successfully treated with oral antibiotic syrup. Infection does not usually become chronic although this may occur and repeated courses of antibiotic treatment may be required. Another organism called *Pseudomonas aeruginosa* is the most common cause of lung infection in older children and adults. However, infection with this organism can occur at any age, even in the early years of life. Aggressive antibiotic treatment can successfully clear *P. aeruginosa* infection in children. However, as children under the age of 7-8 years rarely cough up sputum, your CF doctor may recommend a flexible bronchoscopy and lung wash culture if *P. aeruginosa* infection is suspected. If this bug is found in the lung wash culture, your child's CF doctor will recommend admission to hospital for intravenous antibiotic treatment to clear the infection. If the infection with *P. aeruginosa* becomes chronic, eradication is impossible, even with high dose intravenous antibiotics. Treatment at this stage can therefore only suppress infection, rather than clear it completely. Over the course of months and years, chronic *P. aeruginosa* infection is associated with progressive lung damage and reduced lung function.

### **Monitoring Lung Disease in Cystic Fibrosis**

As mentioned previously, the lungs of newborn babies with CF are normal at birth. However, over the course of months and years, recurrent infections lead to progressive lung damage. Deterioration in lung function occurs very slowly, and can be difficult for parents to detect. We therefore recommend regular out-patient follow-up to ensure prompt detection of the earliest changes of CF lung disease. During out-patient clinic visits, your child will be seen by the doctor but also by the CF team physiotherapist and dietitian. In the first year of life, out-patient visits are scheduled very 1-2 months. In older children and adults, visits occur every 2-3 months.

In addition to regular review of your child by the CF team, we recommend a yearly chest X-ray, to detect early changes of CF lung disease.

A lung function test (also known as a breathing test or spirometry) is one of the most important ways in which we can follow the progression of lung damage in CF. Children of approximately 5 years and older can usually co-operate with testing and thus provide reliable results. The breathing test measures the FEV<sub>1</sub> (the Forced Expiratory Volume in 1 second). This is the maximum volume of air in litres that a person can breathe out ("expire") in 1 second. If the lung airways are wide open (as would be the case in a person not affected by CF), then a large volume of air can be expired. In CF, the volume of air that can be expired may be reduced if thick and sticky airway secretions are present in the airways. So the FEV<sub>1</sub> can be used as a measure of how blocked the airways are. Thus, the more blocked the airways are, the lower is the FEV<sub>1</sub> and the more severe is the lung disease. On the other hand, a normal FEV<sub>1</sub> for age is a reassuring measure of clear airways and more normal lungs.

### **Pulmozyme**

Older children and adults with CF often have more frequent episodes of infection compared to babies and infants. A small proportion of older people with CF have chronic infection producing a daily productive cough. In this situation, the lower airway secretions become thick, sticky and hard to cough up. An inhaled medication known as Pulmozyme can be useful in helping to clear these secretions. Pulmozyme breaks down DNA, which is present in high concentrations in CF sputum. This action causes the sputum to become thinner and easier to clear with physiotherapy, thus helping to prevent or delay ongoing lung damage due to chronically blocked airways. Your child's CF doctor may recommend Pulmozyme if your child is showing signs of CF lung disease such as a chronic productive cough, recurrent hospitalisations for "tune-ups", persistent *P. aeruginosa* infection or low lung function (see later). If your doctor prescribes Pulmozyme (or any other inhaled medication) be sure to speak with your Physiotherapist regarding necessary equipment and appropriate timing of inhaled medication.

## How does Cystic Fibrosis affect digestion?

Approximately 85% of newly diagnosed babies with CF are unable to fully digest the food that they eat due to problems with their pancreas. The pancreas is a gland that sits behind the stomach. One of the roles of the pancreas is to make digestive enzymes that break down the food that we eat. Normally, these enzymes travel through small tubes in the pancreas (pancreatic ducts) and into the small intestine.

In the majority of children with CF the pancreatic ducts are blocked which prevents the digestive enzymes from reaching the small intestine and therefore food cannot be digested. This is called malabsorption. Malabsorption results in poor weight gain and loose frequent bowel actions which contain undigested fat. We will examine stool samples from your baby to look for malabsorption. A large amount of fat in the stool confirms malabsorption and therefore the presence of **pancreatic insufficiency**. Pancreatic insufficiency is treated by giving your baby a pancreatic enzyme extract before every feed. We typically refer to these extracts as “enzymes”.

### Enzymes

Enzymes are extracted from pig pancreases. These enzymes are designed to resist the acid environment of the stomach so they can make their way to the small intestine where they help to digest food. Enzymes for babies come in the form of granules. These enzyme granules are administered mixed with apple puree on a spoon. Our CF Dietitians will show you how to do this.

It is very important that enzymes are taken with each meal, snack or feed to ensure normal digestion and weight gain. For babies, enzymes are given immediately before every breast or bottle feed. If the feed lasts longer than 30 minutes, a second enzyme dose may be required. Your child’s CF Doctor and Dietitian will advise on the initial enzyme dose. Sometimes the dose needs to be altered, to achieve normal weight gain. It is important to tell your Maternal and Child Health Nurse that your child has been diagnosed with CF so visits for regular weight checks can be arranged if required.

### Fat Soluble Vitamins

Absorption of fat-soluble vitamins in the diet is poor in babies with CF who are pancreatic insufficient. Often your child will be placed on a vitamin supplement after diagnosis. Your CF Doctor or Dietitian will recommend an appropriate vitamin supplement.

### Electrolyte Powder

All children with CF lose more salt in the sweat than unaffected individuals. We therefore recommend a daily dose of electrolyte (salt) powder in all babies with CF. Electrolyte powder is given for the first few years of life, until children with CF are old enough to add salt to their meals or take salt tablets. Your child’s CF Doctor and Dietitian will advise you on the electrolyte dose required.

### Dietary Recommendations in Cystic Fibrosis

The recommendations for how to feed your baby do not change with the diagnosis of CF. We recommend that all babies with CF should be breast fed, if possible. If breast feeding is not possible, any of the currently available infant formula feeds are suitable. If stool analysis shows malabsorption, enzymes will be required to ensure adequate digestion of breast milk or formula. As per normal recommendations solids should be introduced when your child shows signs of being ready (at around 6 months and not before 4 months). Our CF Dietitians will assist you with the introduction of solids and enzymes for these solids. Infants with CF sometimes require a diet that is relatively high in fat. This is because even with enzymes,

the digestion of fat is not as good as that achieved by a normally functioning pancreas. Our CF Dietitians will help you provide your child with the appropriate diet and enzymes to meet their individual needs.

## **How does Cystic Fibrosis affect the liver?**

The liver is sometimes affected by CF. The most commonly seen abnormality is an increase in liver enzyme levels. This is detected with a routine yearly blood test. In babies and children, the increase in liver enzymes in the blood is mild, and children with CF have no symptoms of liver disease. However, a small number of older children and adults can develop significant CF liver disease, producing enlargement of the liver and other symptoms such as abdominal distension, as well as enlargement of the spleen. A very small number of adults with CF develop progressive liver damage (or liver cirrhosis). This can sometimes be managed with medication, but if liver failure occurs, liver transplantation is required.

## **Family and Parenting perspective**

**During the first week after diagnosis**, many couples find it useful to take the time to discuss their feelings and questions with their partner, friends or family members. It's important that parents are able to talk things through with each other and with the CF team. It is also important to remember that the social worker is happy to meet with you and your family to discuss any concerns. The CF team is committed to on-going education and support, which starts when your baby has been diagnosed. During this first week, it is important for both parents to attend where possible.

During this period of diagnosis, parents may experience many different feelings inclusive of disbelief, anxiety, confusion, stress and anger. It is important you to know that these feelings are normal and decrease with time. Feedback from other families tell us that they experience many different emotions and that the more familiar and comfortable they became with CF information and the daily treatment routine - the more confident, relaxed and less worried they felt about managing their feelings and the more optimistic they felt about managing the illness. For some families feelings of relief are experienced, especially when their baby has been unwell or unsettled before a diagnosis.

## **Coping with the challenges of caring**

The responsibility of caring for a new baby is demanding and the special needs of a baby with CF may seem overwhelming; besides normal parenting challenges, parents of children with CF must attend to physiotherapy, medication, enzymes, exercise and sometimes, hospitalizations. The aim of the CF team is to work with your family in supporting and strengthening your own existing parenting skills in working with your child's CF diagnosis. We are also committed in supporting your growing child's independence and self-reliance in managing their health. Our team will continue to work with you and your growing child through their childhood, adolescence and adulthood.

The team also works closely with maternal and health nurses, kindergarten, primary, secondary teachers, university staff, GP's, Cystic Fibrosis Victoria (CFV) and other community organizations in supporting their knowledge of Cystic Fibrosis. Monash CF also holds regular education days during the year for parents, their families and friends. CFV provides support programs and activities and networks, information, financial assistance for physiotherapy equipment and other costs and a comprehensive CF information handbook if you join CFV.

## **Family and Friends**

Most of the time, friends and family provide the best support. You will probably know who will be most supportive and helpful during this time. At the time of a new diagnosis, families may however receive

well-intended but misinformed advice or information from others. This can be stressful for parents and if you do feel unclear in relation to information you have heard, it is always useful to check in with the team until you feel confident about any aspects of CF care. Although the internet is a useful tool, it is best to ask the team which is the best informed and up-to-date websites - to avoid out of date or incorrect information. The social worker can support you and discuss any concerns you may have.

## **Siblings**

If you have other children, it is common for children in the family to start 'acting out' after the birth of another baby. These normal developmental responses by a sibling may present an extra challenge during the period of a new diagnosis. It is common for children to test or push boundaries especially when you are naturally more focused on the needs of your new baby. Given a return to family routine, soon after diagnosis, these behaviours usually reduce and it is useful to retain the normal patterns and limits that were in place beforehand. Siblings can be encouraged in everyday treatment routines and exercise. Encourage your child and family members to talk openly about CF with friends, family, class and workmates.

### **How do we know if siblings have C.F.?**

Other siblings can be tested here at Monash Medical Centre, which involves a simple and painless test, known as a 'sweat test.' This measures the level of salt (sodium) in your child's sweat. A positive sweat test indicates a significant increase in sodium levels and the diagnosis of CF.

### **Can Cystic Fibrosis be diagnosed before birth?**

Yes, testing can be carried out at Monash Medical Centre at 10-11 weeks of pregnancy and genetic counselling is available to all family members.

Families often say, when talking about their experience of a CF diagnosis, was the importance of taking on information at your own pace. Some families will read all this material within the first couple of days. Others may choose to read very little during the period of diagnosis, preferring to discuss most aspects of care with the team, before feeling ready to read any written material. There is no right or wrong approach and the team will support your family's particular learning style, which feels most comfortable for you.

## **What Families Say: feedback a year after diagnosis**

*'I found the first few days difficult, but the more I learnt about CF and the more I spoke to the CF team, the more reassured and confident I felt.'*

*'Once I learnt that my child was able to achieve all the things that other children achieve (going to school, playing sport, going to uni, getting married and having a child), I was able to deal with things more positively.'*

*'The team was very optimistic about the future of gene therapy and present research and I found that very reassuring.'*

*'Looking back on that time of diagnosis, I realize how important it was to take things in my own stride and really use all the resources offered by the CF team.'*

*'I found it helpful to ask lots of questions, often asking the same questions over and over, until the answers somehow stuck. The team was very supportive and were happy to spend as much time as I needed for things to make sense.'*

*'I have come to realize that CF is just one small part of my child's developing life.'*

## Summary

With modern treatment regimes, we expect that all newly diagnosed CF infants will lead productive healthy lives well into adulthood. Whilst your child has CF, it does not stop him or her from participating fully in life. The Monash CF team will work with you to provide the best care for your child. As parents, we ask you to encourage your child to

- Have a positive and optimistic approach to life
- Learn an early sense of independence, understanding of CF and responsibility for self-care in partnership with his/her CF team
- Develop positive self-esteem and coping strategies
- Lead a social and active lifestyle
- Complete his/her education and progress to further study and employment
- Plan for his/her future in the same way as siblings and friends

## Acknowledgements

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