CFstrong
Inform. Educate. Empower.

A Guide for Health Professionals

Contents

Cystic Fibrosis Overview

Cystic fibrosis (CF) is a multi-organ life shortening disease typified by chronic endobronchial infection and progressive obstructive lung disease and malnutrition, secondary to pancreatic insufficiency. CF is the most common autosomal recessive condition in Australia, affecting approximately one in every 3,700 babies born in this country. Previously a disease of childhood, however, with advances in clinical care, currently there are more adults with CF than children.

While there is no cure as yet, the introduction of mutation-specific therapies and specialised multi-disciplinary treatment has led to an impressive increase in survival in recent decades. It is projected that children born in the UK in 2018 can be expected to live beyond 50 years, as well as those living with CF today who are 30 years or older (Keogh, Szczesniak, Taylor-Robinson, Bilton, 2018).

With newborn screening, many CF centres worldwide are now caring for children with minimal lung damage. It is encouraging that those with CF have the potential to enjoy an increasing life span and an excellent quality of life well into adulthood.

Although life expectancy has improved greatly, the majority of people with CF still die of respiratory failure. The primary aim of CF therapy is to slow the progression of lung disease.

Additional CF treatment goals include:

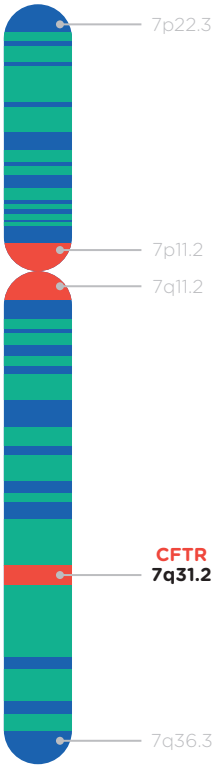
- optimal nutrition and management of metabolic complications
- treating complications of CF in a timely way
- reducing early bacterial colonisation
- psychosocial support
- slowing airway inflammation to preserve lung function
- avoiding pulmonary exacerbations
- attainment of normal growth in childhood
- maintenance of adequate nutrition in adulthood
- transplantation and appropriate management of end-of-life care.

The CF Gene

The gene for CF results from a mutation in the cystic fibrosis transmembrane conductance regulator (CFTR) gene located on chromosome seven. Over 2,000 CFTR mutations have been discovered; many of these are known to cause CF. The variation in signs and symptoms are associated with the specific CFTR variant or genotype. Genetic background and various environmental factors influence clinical outcome. In healthy individuals, the CFTR protein forms within the cells and travels to the membrane. It then acts as a channel allowing chloride ions to flow out of the cell.

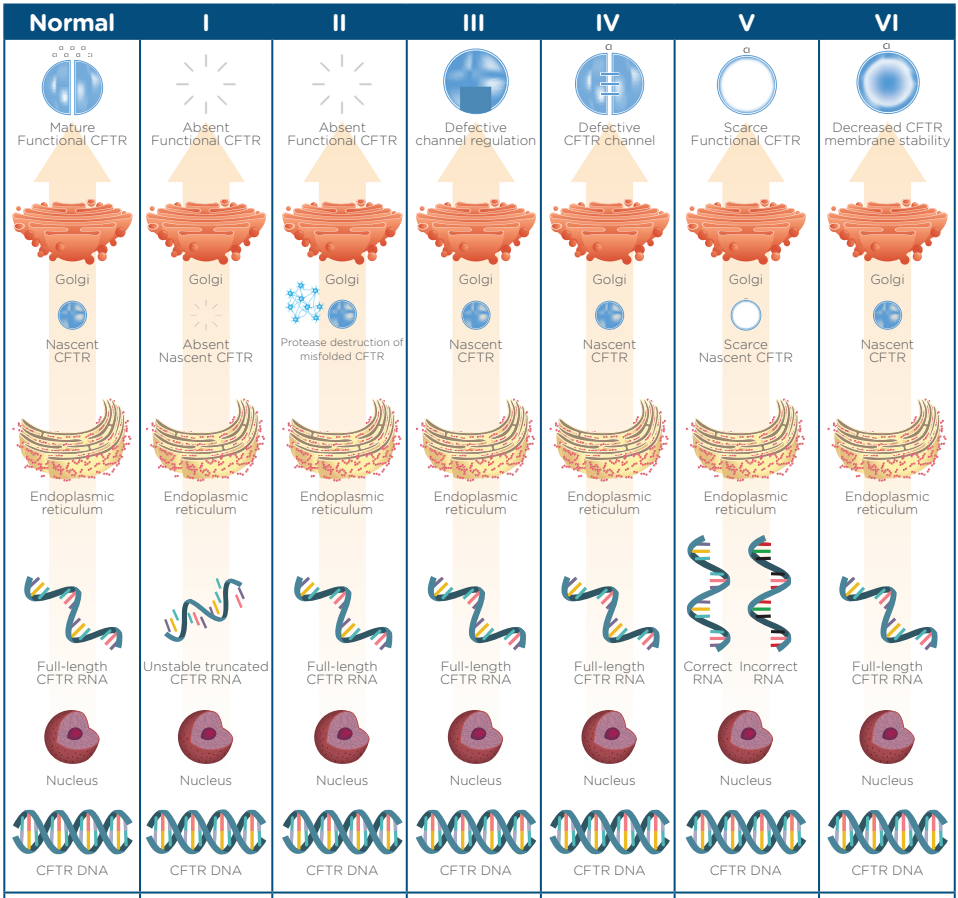
Disease causing mutations of the CFTR DNA code result in either a lower amount, or an alteration, that leads to a reduction in the CFTR that reaches the cell surface. This CFTR protein reduction means there is decreased chloride secretion and increased sodium absorption in epithelial cells. This causes impaired movement of water in and out of the cells, resulting in abnormal mucus production in the lungs, pancreas, gut and other organs. The alteration in airway surface liquid (ASL), and the resultant increase in viscosity compromises mucociliary clearance, resulting in a predisposition to pathogen colonisation.

> *CF is most common amongst the caucasian race*



Chromosome 7

The CF Gene Mutation Classification



Normal	I	II	III	IV	V	VI
CTFR Defect	No Functional CTFR Protein	CTFR Trafficking defect	Defecting channel regulation	Decreased channel conductance	Reduced synthesis of CTFR	Decreased CTFR stability
Type of mutations	Nonsense; frameshift; canonical splice	Missense; aminoacid deletion	Missense; aminoacid change	Missense; aminoacid change	Splicing defect; missense	Missense; aminoacid change
Specific mutation examples[11]	Gly542X Trp1282X Arg553X 621+1G→T	PheS08del Asn1303Lys Ile507del ArgS60Thr	GlySSIAsp Gly178Arg Gly551Ser Ser549Asn	Arg117His Arg347Pro Arg117Cys Arg334Trp	3849+10kbC→T 2789+SG→A 3120+1G→A 5T	4326delTC Gln1412X 4279insA

(Boyle & De Boek, 2013)

Newborn Screening

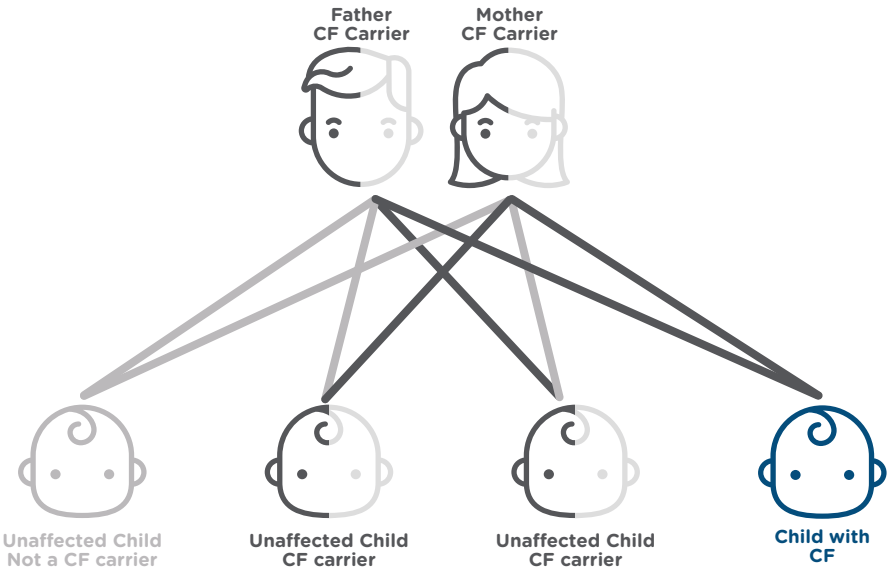
In Australia, cystic fibrosis was added to the newborn screening (NBS) test in 1981 in NSW, and was eventually adopted by all states and territories by 2001.

CF is a common inherited disorder with birth prevalence in Australia reported to be approximately one in 3,700 births (Ruseckaite et al, 2016). Around one in 25 Australians are carriers of a genetic mutation responsible for CF.

Children must inherit two defective CFTR genes – one from each parent – to have CF. It is important for those with a family history of CF to be aware of the options for carrier screening. Newborn screening is not the same as carrier screening. Only a very small proportion of CF carriers are identified by newborn screening.

Newborn screening for CF is a two-step process from the heel prick test at birth. It is mainly targeted at detecting elevated levels of immunoreactive trypsinogen (IRT) in the newborn's blood. Mucus plugging in the pancreatic ducts in a newborn with CF may cause blockages that prevent trypsinogen from reaching the small intestine. This results in an elevated serum trypsinogen. A positive IRT is followed by DNA testing to screen for the most common CF genetic mutations. Infants with one or two common mutations are usually recalled for a sweat test at four-to-six weeks of age.

The most commonly known mutation in CF is delta F508.



Carrier Screening

Carrier screening is not the same as newborn screening. Only a very small proportion of CF carriers are identified by newborn screening. It is important for those with a family history of CF to be aware of the options for carrier screening, in order to make informed decisions associated with family planning.

Cystic Fibrosis Community Care Carrier Screening Program: www.cysticfibrosis.org.au/get-involved/support-and-services/support-and-services-information/cf-carrier-screening

60-75
people

are diagnosed with CF each year in Australia. *(Approximately)*

1
Person

in 25 carries the gene for CF.

> *Most screening programs screen for CF along with phenylketonuria, galactosaemia, congenital hypothyroidism, and a range of disorders of amino, organic and fatty acid metabolism.*

Clinical Manifestations and Management

Although CF is a multi-organ disease, it is the pulmonary disease that presents the most challenge and continues to be the main cause of mortality. Most people with CF have respiratory disease and exocrine pancreatic insufficiency. The CFTR defect is expressed in many epithelial cells including sweat ducts, airway epithelium, pancreatic ducts, intestine, biliary tree and vas deferens.

Clinical manifestations may include:

Respiratory

- Cough
- Increased purulent sputum
- Dyspnoea, hypoxia, tachycardia
- Bronchiectasis
- Haemoptysis
- Lung function decline
- Chest pain
- Pneumothorax
- Sinusitis
- Nasal polyps
- Lethargy/fatigue
- Fever

Gastrointestinal

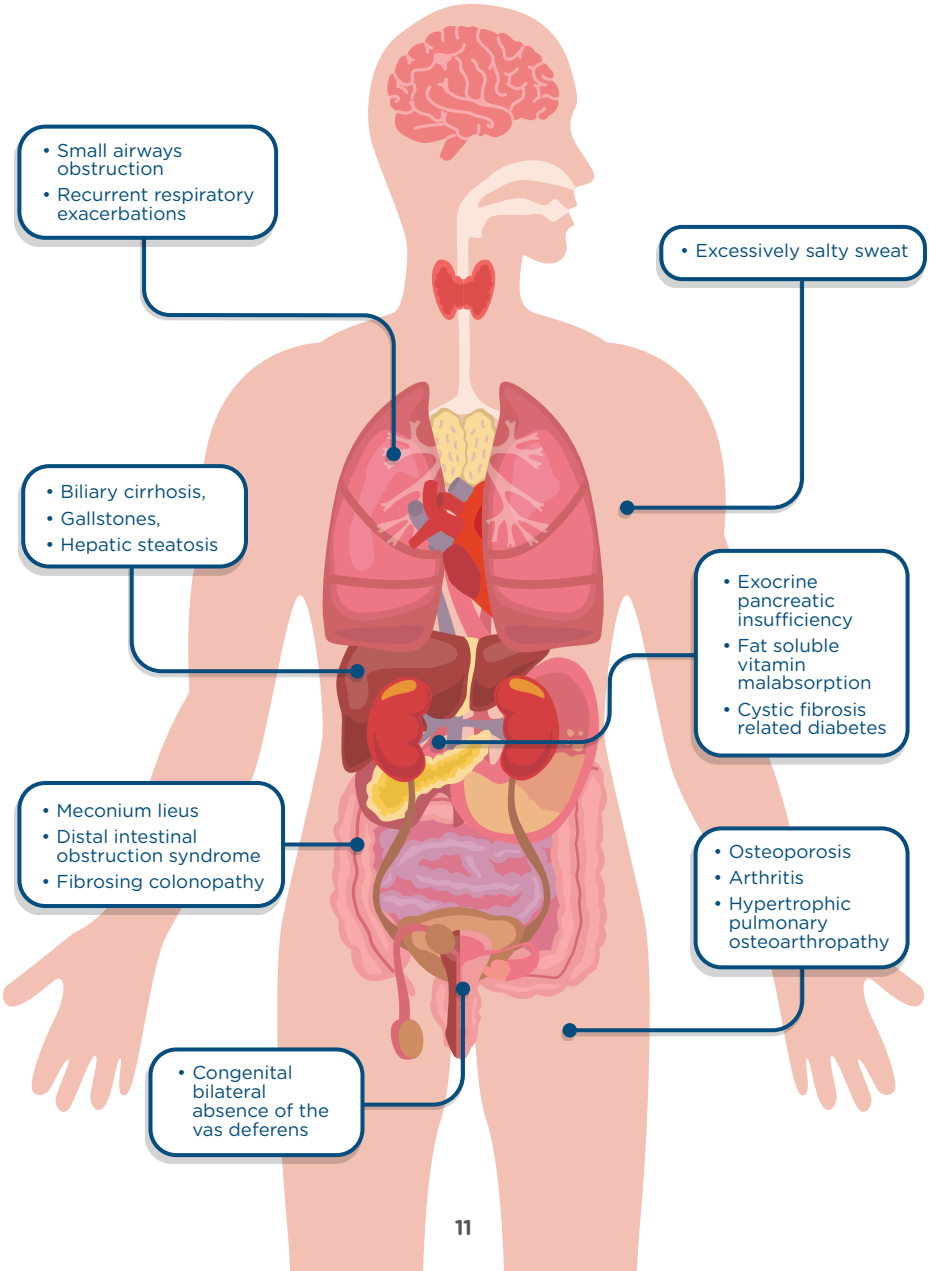
- Pancreatic insufficiency
- Loss of appetite/weight loss
- Abdominal pain
- Gastro-oesophageal reflux disease (GORD)
- Pancreatitis
- CF-related diabetes (CFRD)
- Rectal prolapse
- Distal intestinal obstruction syndrome (DIOS)
- Cirrhosis and other hepatic dysfunction
- Cholelithiasis
- Dehydration

Psychosocial

- Anxiety and depression
- Body image disorder

Other

- Bone and joint disease including osteopenia and osteoporosis
- Genitourinary disease including infertility in males and stress incontinence
- Kidney disease

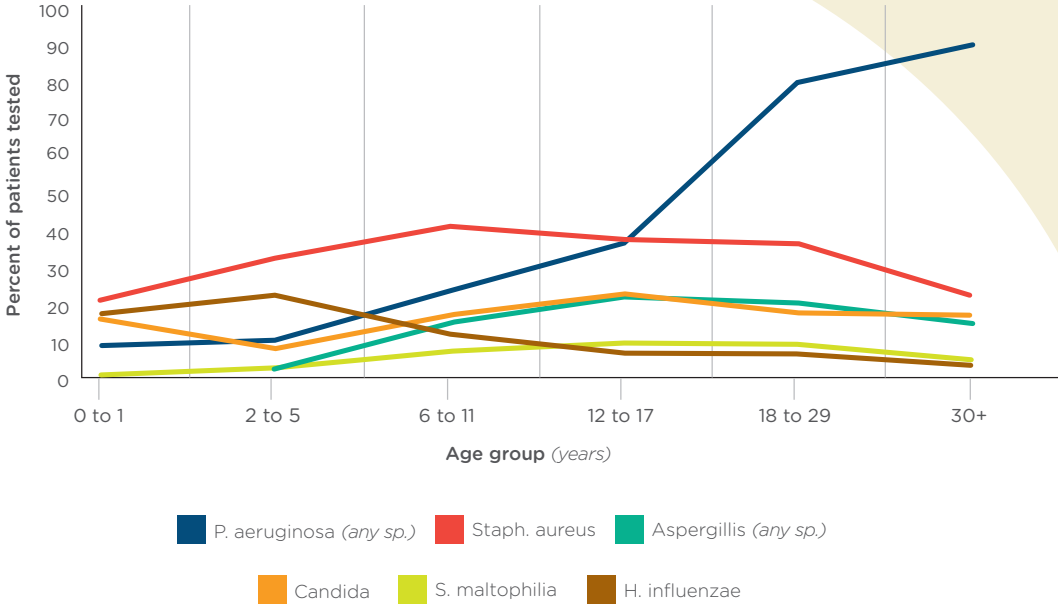


Respiratory

The chloride and sodium channel defect results in thicker more viscous secretions from exocrine glands, particularly in the respiratory tract, and results in extensive chronic inflammation of the airways. The recurrent lower respiratory tract infections, and a chronic cough with sputum production, leads to structural airway changes including bronchiectasis, obstructive lung disease, and finally, respiratory failure. Bronchiectasis develops in infancy in cystic fibrosis *(CF)* and may be asymptomatic. Studies have shown that neutrophilic inflammation and pulmonary infection, especially *Pseudomonas aeruginosa (Pa),* are major risk factors for early disease in CF, including nutritional and lung function decline *(Sly et al, 2013).*

The Australian CF Data Registry *(Ruseckaite et al, 2017)* describes the prevalence of respiratory infections and reports that in childhood these are predominantly related to *Staphylococcus aureus* and *Haemophilus influenza,* with Pa dominating adult years.

Prevalence of Major Organisms in Lungs



(Ruseckaite et al, 2017)

A comprehensive approach to maintaining lung health includes airway clearance, physical activity, drug therapy and optimal nutrition.

CF care becomes more complex with increasing age and a multidisciplinary health care team is essential. The focus for pulmonary management in CF relates to prevention of airway obstruction by improving mucociliary clearance and prevention and early treatment of respiratory tract infections. A comprehensive approach to maintaining lung health includes airway clearance, physical activity, drug therapy and optimal nutrition.

Airway Clearance

Airway clearance is an important part of CF management and should be used across the lifespan. The physiotherapist's role involves teaching effective clearance of secretions from the lungs and promoting optimal lung health and general physical fitness. Research has found that no single airway clearance technique is superior, and since CF affects each person differently, a treatment program is designed specifically for the individual and needs to be reviewed regularly. Regular contact with the physiotherapist can help identify small changes, which may be hard for the individual to detect, thus allowing adjustments to the treatment program.

Physiotherapy treatment should be increased when a lung infection occurs and may be more effective when combined with inhaled medications and exercise *(Button et al, 2016)*.

Modified Postural Drainage

For babies and young children, when active participation is not possible, the basic program usually involves daily or twice daily modified postural drainage *(MPD)* with percussion. MPD involves positioning without the use of a head-down tilt. As children grow, physio 'play', blowing games, coughing and deep breathing exercises are incorporated into the treatment.

Active Cycle of Breathing Technique

From about three to five years of age, more emphasis is placed on breathing exercises. The active cycle of breathing techniques *(ACBT)* which incorporates deep breaths, breathing control and the forced expiration technique *(FET),* is taught as a method of clearing mucus more independently. Studies have shown it to be an effective method of mobilising and clearing secretions.

Positive Expiratory Pressure

Devices such as the positive expiratory pressure (PEP) mask, mouthpiece PEP, bottle PEP, Flutter or Acapella may be prescribed by the physiotherapist to enhance airway clearance. PEP therapy is thought to promote collateral ventilation, increase functional residual capacity and recruit obstructed or collapsed airways.

PEP increases the volume of air behind an obstruction and creates pressure to force secretions centrally, towards the larger areas, where they can be more easily cleared. Bottle PEP can be used to help transition younger children to a PEP device or may be used in hospital where there is no access to expensive devices. PEP can also be used in babies and toddlers when appropriate.

Autogenic Drainage

Autogenic Drainage *(AD),* or self-drainage, is an airway clearance technique that is widely used throughout Europe. The technique is based on the principle of reaching the highest possible airflow in different generations of bronchi by controlling breathing. When performing AD, individuals adjust the rate, depth and location of respiration in order to mobilise secretions.

Physical Activity

Another very important early recommendation for airway maintenance is regular exercise. Physical exercise that increases minute ventilation leads to the mobilisation of secretions and enhances airway clearance. It is recommended that people with CF should all be encouraged to exercise several times a week.

Drug Therapy

Managing CF is complex and drug therapy is an important part of CF treatment. Medications are used to ease symptoms, reduce complications and improve quality of life. They may be inhaled, oral or given parentally. A major achievement in treatment are medications that target gene mutations known as CFTR modulator therapies.

Central Vascular Access Devices

Those with CF may require long term antibiotics to treat respiratory exacerbations. Peripherally Inserted Central Catheters *(PICC)* are used when vascular access is required for more than a week. Infusaports are used for long term access.

Mucolytics

Inhaled mucolytic agents are often used as an adjunct to airway clearance techniques and allow direct deposition of drugs to reduce mucus viscosity.

Dornase alpha *(Pulmozyme®)*

Pulmozyme® is a recombinant human deoxyribonuclease *(rhDNase).* In those with CF, Dornase alpha hydrolyses the DNA of sputum and reduces the viscosity, enhancing airway clearance with the potential to increase lung function and reduce exacerbations.

Bronchitol® and inhaled hypertonic saline (HTS)

Bronchitol® and HTS are both examples of hydrator therapies. Accelerated sodium transport at the cellular level dehydrates the airway and impairs mucus clearance. Hydrator therapies are used to restore airway surface liquid hydration by osmotically drawing liquid into airways. Most people will require a bronchodilator, such as Salbutamol®, before inhaling Bronchitol® or HTS.

Bronchitol® is a form of mannitol, a naturally occurring osmotic agent. It is delivered via a dry powder inhaler prior to airway clearance.

Inhaled HTS has been shown to increase mucociliary clearance and improve clinical outcomes in children and adults with CF. It is recommended that HTS is administered via a nebuliser before or during airway clearance.

Antimicrobial Therapy

Antimicrobial therapy has been strongly associated with increased survival in those with CF. Long-term use of antibiotics has been significant in slowing lung decline.

Azithromycin is often prescribed for children in subtherapeutic doses. These macrolides are thought to suppress proinflammatory cytokines and reduce the neutrophil burden on the lungs.

Tobramycin inhaled solution *(TOBI®)* or inhaled powder *(TOBI Podhaler®)* – the introduction of inhaled Tobraymicin® was a significant milestone in CF therapy. It is indicated for the management of Pseudomonas aeruginosa.

Modulator Therapy

CFTR modulators are a new drug therapy that differ from other CF therapies as they aim to improve or restore the function of the defective CFTR protein, rather than offer symptomatic treatment. These therapies are mutation specific.

The individual modulators act in different ways to improve the mutant protein function by potentiating action at the cell surface (increasing CFTR channel opening) or correcting the defect by increasing the amount of CFTR protein at the cell surface. Work continues on new modulators in an effort to find an effective treatment for all people with CF.

Kalydeco®

Kalydeco® is used in people with at least one mutation that is responsive to Kalydeco®, including the G551D gating mutation. Kalydeco® binds to the defective protein, potentiating the channel opening (gating) of the CFTR protein. Chloride can flow through and regulate the amount of water and cell level.

Orkambi®

Orkambi® is a combination corrector therapy for those with two copies of the F508 del mutation. It facilitates increased chloride transport by helping the CFTR protein to become the right shape to get to the cell membrane. Used with Kalydeco®, the gate is opened, and more chloride flows through and reduces the symptoms of CF.

Symdeko®

Symdeko® is a combination of Tezacaftor® and Orkambi®. Patients must have two copies of F508 del or at least one mutation that is responsive to Symdeko. Tezacaftor® facilitates the cellular and processing and trafficking of normal and mutant forms of CFTR which increases the amount of mature CFTR delivered to the cell surface. Orkambi® can potentiate the CFTR to the cell surface by Tezacaftor®, increasing the quantity and function of the CFTR at the cell surface and improving the chloride transport.

Trikafta®

Trikafta® is a next-generation CFTR corrector designed to restore F508 del CFTR function. It is used for the treatment of people with CF who have at least one copy of the F508del mutation in the CFTR. Trikafta® is currently not listed on the Australian Register of Therapeutic Goods (ARTG).

Respiratory Complications

While progressive bronchiectasis and recurrent pulmonary exacerbations are typical of CF lung disease, additional respiratory complications also occur.

Pneumothorax

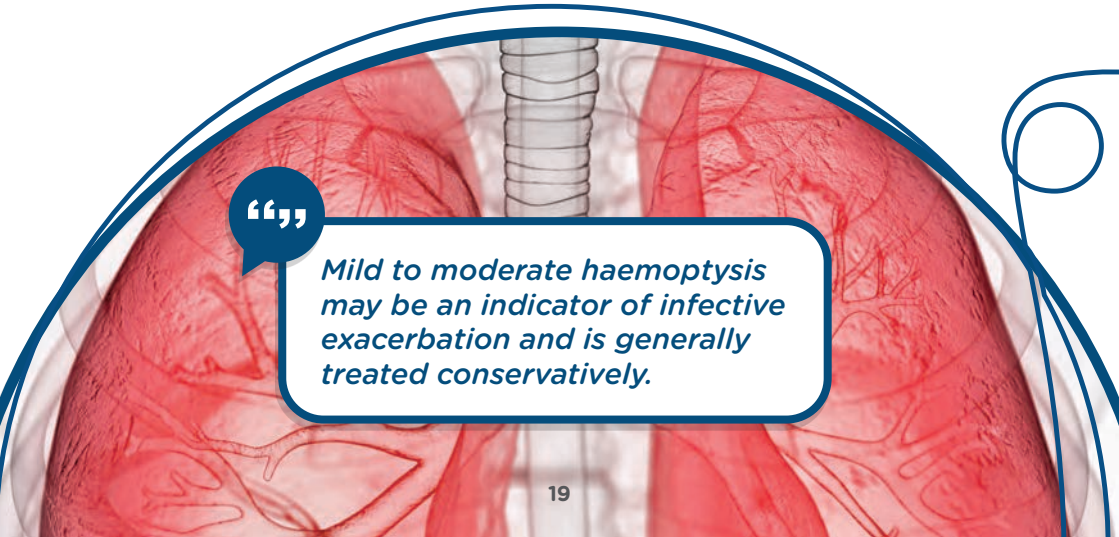
Pneumothorax is defined as the presence of air or gas in the space between the visceral and parietal pleura of the lung, which can impair oxygenation and/or ventilation. A sudden onset of breathlessness in people with CF should be investigated promptly to identify the possibility of a pneumothorax. In more severe cases, hypoxemia and hypercapnia may be observed *(Ronan, Elborn, Plant, 2017).*

In CF, pneumothorax is a marker of disease severity and often associated with lower FEV1, pancreatic insufficiency, haemoptysis and infection. While a small pneumothorax can be managed conservatively, a larger pneumothorax may require surgical intervention.

Haemoptysis

Haemoptysis is defined as the expectoration of blood, or blood-stained mucus, from the lower respiratory tract.

Mild to moderate haemoptysis may be an indicator of infective exacerbation and is generally treated conservatively, withholding some of the usual CF treatments and adding antibiotics to treat infections. Treatment of moderate haemoptysis may also include tranexamic acid. Massive haemoptysis may be associated with a positive sputum culture for *Staphylococcus aureus* and cystic fibrosis related diabetes *(CFRD).* It is life-threatening and often requires surgical management *(Ronan, et al, 2017).*



> ***Mild to moderate haemoptysis may be an indicator of infective exacerbation and is generally treated conservatively.***

Gastrointestinal

Patients with CF may present at birth with meconium ileus and have higher incidence of complete or partial bowel obstruction, commonly known as DIOS. They may also be more prone to constipation, gastro-oesophageal reflux disease, and infections such as C. Difficile. Approximately 90% of Australian CF patients are pancreatic insufficient with consequential poor weight gain, malabsorption of fat and fat-soluble vitamins *(Ronan et al, 2017).*

Lung function and nutritional status are closely linked. Malnutrition related to inadequate energy intake is a common problem in CF patients. Interdisciplinary management is essential with involvement of a dietitian, gastroenterologist and endocrinologist to manage gastrointestinal *(GI)* co-morbidities.

Pancreatic Insufficiency

Pancreatic insufficiency refers to significant impairment of the pancreas to secrete sufficient enzymes needed for normal digestion *(Saxby, et al 2017).* In individuals with CF, the exocrine glands in the pancreas produce such thick secretions that the pancreatic ducts become blocked. Destruction of acinar pancreatic tissues occurs and lack of enzyme activity results in malabsorption, particularly of fatty foods and fat-soluble vitamins.

The resultant malnutrition and poor growth associated with fat malabsorption and pancreatic insufficiency is often accompanied by steatorrhea, abdominal pain and failure to thrive. With CFTR modulator therapy, the emergence of the overweight person with CF is an increasing phenomenon. It is important that people are screened for undernutrition and overnutrition.

Nutritional Recommendations

The energy requirements associated with malabsorption, increased work of breathing, chronic inflammation and infection result in recommendations of an energy intake 120%- 150% greater than the general population. A diet unrestricted in healthy fats and high in carbohydrates leads to better growth. Recommendations include:

- pancreatic enzyme replacement therapy (PERT) for those with pancreatic insufficiency
- high energy/high fat diet to optimise weight for those undernourished (healthy fats are encouraged)
- sodium supplementation
- supplementation with fat soluble vitamins (ABDECK®)
- nutritional supplements may be required to optimise nutritional status
- enteral feeding if indicated
- use of proton pump inhibitors (PPI) for the treatment of gastro-oesophageal reflux disease (GORD)
- behavioural therapy to achieve positive meals times

General Population Food Pyramid

CF Population Food Cube

Pancreatic Enzyme Replacement Therapy *(PERT)*

Pancreatic enzyme replacement therapy *(PERT)*, along with vitamin supplementation, is an essential part of the management of pancreatic insufficiency in CF *(Saxby et al, 2017).* It is important that pancreatic enzyme replacement capsules are taken with the first mouthful of foods that contain fat, carbohydrates and protein. The enzyme dosage is related to the fat content of food ingested and an additional dose may be needed if the meal lasts more than 30 minutes.

Creon® is a commonly used PERT in CF. It has a slow release formula designed to deliver enzymes to the duodenum. It can be given as granules mixed with an acidic fruit puree in infants and small children or taken as capsules in older children and adults. PERT adequacy is assessed clinically by monitoring nutritional status, signs of malabsorption and weight gain.

Supplements

Vitamins

Those with CF, especially the pancreatic insufficient, are at risk of fat-soluble vitamin deficiencies. VitABDECK® is a CF specific multivitamin for routine supplementation.

Sodium

The CFTR defect results in abolition of normal chloride conductance with consequently poor sodium reabsorption into the cells and a high concentration of salt in sweat. This increased loss of sodium and chloride with exercise and in hot temperatures, predisposes the person with CF to salt depletion and dehydration. Sodium supplementation may be recommended

Enteral Feeding and Nutritional Supplements

Enteral feeding can be an effective way to optimise nutrition in those not meeting nutritional requirements. Low profile Percutaneous Endoscopic Gastrostomy *(PEG)* tubes are often used in children. Intermittent nasogastric tubes may be used by those who would rather a less invasive approach. The CF dietitian will prescribe the appropriate nutritional supplements.

Gastrointestinal Complications

The most commonly presenting gastrointestinal complications are cystic fibrosis related diabetes *(CFRD)* gastro-oesophageal reflux disease, liver cirrhosis, portal hypertension, pancreatitis, cholelithiasis, constipation, distal intestinal obstruction syndrome *(DIOS)* and rectal prolapse. Lack of functional CFTR in biliary epithelium results in increased viscosity of bile and may lead to liver complications.

CF Related Diabetes *(CFRD)*

The prevalence of CFRD increases with age and is becoming one of the most common co-morbidities associated with CF. It is linked with decreasing lung function, poor nutrition and increased respiratory exacerbations. Acinar atrophy, fatty infiltration and pancreatic fibrosis occur with advancing age and results in decreases in insulin producing beta cells *(Ornstein, Rosentstein & Stern, 2000).* CFRD affects approximately 20% of adolescents and 40-50% of adults *(Ronan et al, 2017).*

Although CFRD shares similar characteristics with type 1 and type 2 diabetes, it is managed with a typical CF, high energy diet and insulin. All those with CF should be screened for diabetes annually from 10 years of age.

Psychosocial

CF is a chronic life-limiting disease, with a complex and high treatment burden. Anxiety and depression are reported to be two to three times higher for people with CF and their carers *(Quittner, Abbot, Georgiopoulos, et al, 2016).*

This can be exacerbated at times of diagnosis, hospital admission, specific medical interventions or another diagnosis, such as CFRD.

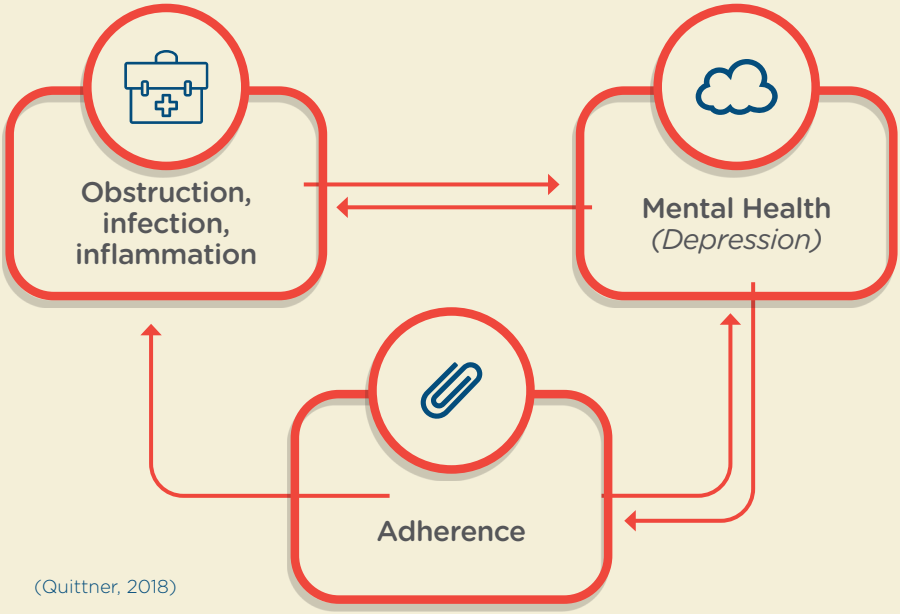
Anxiety and depression are normal at times of stress, however, if untreated can lead to unresolved grief and complex trauma which is associated with poor health literacy, low adherence to treatment regimens and poorer health outcomes.

The incidence of depression among children and adults with CF has been reported to be up to 30%. It is recommended that adolescents and adults with CF *(ages 12yrs – adulthood)* are screened annually for depression and anxiety *(Quittner et al, 2016).*

Living with CF can be emotionally and physically challenging for the person and their family. It is important to recognise and treat anxiety and depressive symptoms in people with CF and their carers, as this can impact on treatment adherence, family functioning and quality of life, and has been associated with reduced lung function and increased hospitalisations.

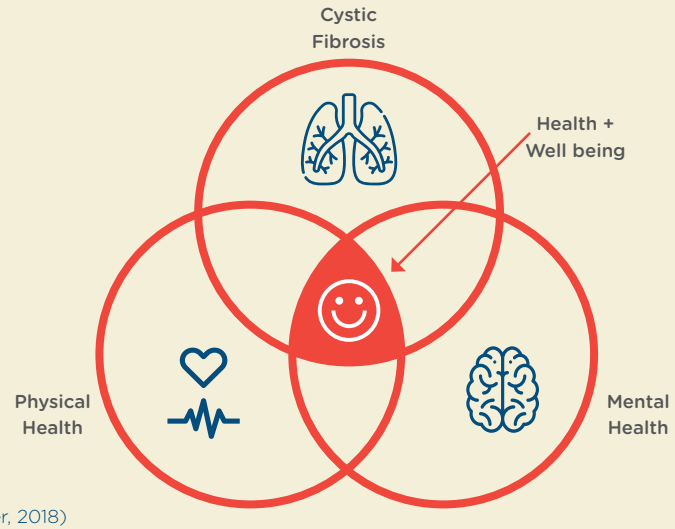
Patients with anxiety and depression are eligible for up to 10 individual counselling session and 10 group counselling sessions per calendar year, which are subsidised by Medicare.

Impacts of Cystic Fibrosis



(Quittner, 2018)

Treating the Whole Person



(Quittner, 2018)

Infection Prevention and Control

Patients with CF are at risk of patient-to-patient and environment-to-patient transmission of respiratory pathogens through droplet, contact or airborne mechanisms. Multi-resistant organisms are a concern and may not be identified until screening or testing is done.

It is recommended that patients with CF maintain a minimum distance of four meters apart at all times to minimise the risk of transmission of micro-organisms. It is important to try to minimise exposure to common respiratory pathogens. Admit patients to a single room when hospitalised.

Annual flu vaccination is also recommended.

Transmission-based precautions for all patients accessing hospital services include:

- allocation of single room with own bathroom
- patients to remain in their room with door closed
- contact precautions for all CF patients, including use of Personal Protective Equipment (PPE)
- all respiratory therapy to be performed in the patient's room with door closed
- single use/patient dedicated equipment
- use of TGA registered disinfectant such as Nocospray/ OxiverTb solution
- patient should wear a mask in common areas including ED, outpatients, pathology and radiology.

Extra precautions for people with CF known to be infected/colonised with Mycobacterium abscesses and Burkholderia cepacia include:

- use of negative pressure single room if available and own bathroom essential
- respiratory function testing to occur in patients own room with portable equipment if possible
- patients should remain in their room and not visit other areas of the hospital
- patients/carers/visitors must clean their hands with alcohol-based hand rub or soap and water on entering and leaving the patient's room

> ***Contact your local tertiary centre for CF patient infection prevention and control management guidelines*** *(details on page 36).*

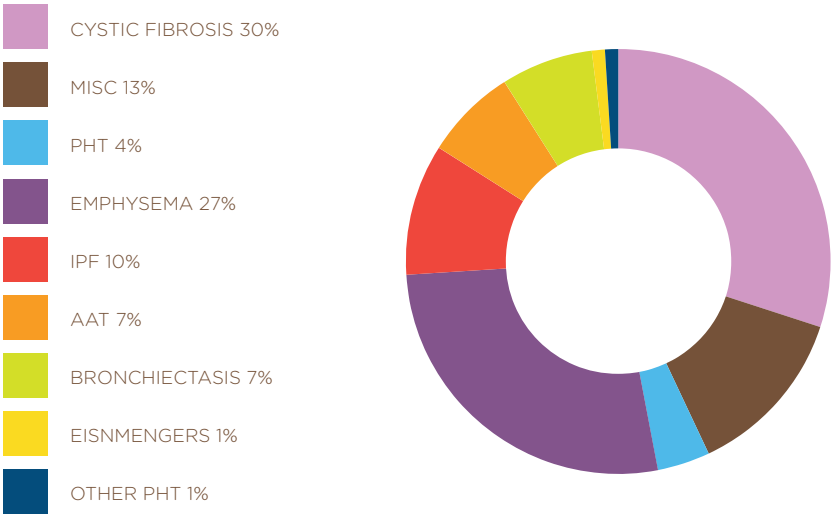
Lung Transplantation

For some people with CF, lung transplantation is an option in managing end-stage lung disease. In Australia, lung transplant programs for those with CF are located in Perth, Brisbane, Melbourne and Sydney. The Paediatric Lung Transplant Program at the Alfred Hospital in Melbourne is the only paediatric service in Australia. Between 1992 and 2015, there were 658 double-lung transplants performed on adults in Australia. In 2016, 51 patients with CF were assessed for organ transplant and twenty-nine bilateral lung transplants were reported by CF centres *(Ruseckalite et al, 2018).* **Australian recipients of lung transplants have a longer survival rate than the global average.**

Guidelines for consideration for transplant in CF include:

- FEV1 falling to 30% predicted or rapidly falling, particularly in females
- 6-minute walk test <400m
- Development of pulmonary hypertension
- Clinical decline presented by:
  - Long term non-invasive ventilation use
  - Increase antibiotic resistance
  - Pnemothorax
  - Haemoptysis

Reason for Bilateral Lung Transplant, 1992 - 2018



(Keogh, Williams & Pettersson, 2018)

51

patients with CF were assessed for organ transplant

29

bilateral lung transplants were reported by CF centres

Family Planning

Women with CF are now surviving into their reproductive years and manage to have successful pregnancies. Lau et al. *(2011),* **concluded that most women had acceptable outcomes from their pregnancies. However, body mass index and lung function were significant predictors of foetal complications. Pregnancy should be planned and involvement of genetic counselling for the couple should be part of the support team.**

Prior planning can help to optimise lung health and weight, which can lead to improved outcomes for mother and infant. Medications should be reviewed preconception so those that may affect foetal development are ceased. The CF pregnancy is considered high risk and should be managed at a tertiary centre.

Most CF males are infertile due to blocked or absent vas deferens and should consider having a semen analysis as part of family planning. Most males with CF produce normal sperm, and with assisted reproductive technologies, are able to have their own biological children.

Cystic Fibrosis Community Care - Carrier Screening Program
www.cfscreening.com.au

End-Of-Life Issues

People living with CF experience a slow decline in lung function and multi-organ complications may occur over a long period. In the prolonged chronic phase of the illness, people undergo intensive regimes of treatment and predicting prognosis is difficult. Lung transplant is not an option for all those with CF. Variable disease progression can mean that the timing of death may be difficult to predict.

Providing optimal end-of-life care may be challenging due to the variation in disease, with dyspnoea, pain, congestion and anxiety being the most prevalent symptoms. Ideally this would be provided by a specialised palliative care team. With CF, active and palliative care usually occurs simultaneously, and in some cases, life-sustaining treatment will occur while awaiting lung transplant.

Advance Care Planning is particularly important for people living with CF because of the unique aspects of the disease. Planning should reflect and respect the individual's decisions as lung disease progresses.

Useful Contact:

Palliative Care Australia

Phone: 02 6232 0700

Email: pca@palliativecare.org.au

Web: palliativecare.org.au

Continuing Care in the Community

Cystic Fibrosis organisations across Australia are funded by generous community donations, philanthropic grants and government to provide in home and community-based support for people with CF and their families.

While the specific services provided may differ for each state or territory, they can include:

- assistance with airway clearance
- assistance in developing early routines
- exercise programs
- transition support for young people and carers
- counselling/general support
- financial support
- face-to-face education for daycares, schools, workplaces and health professionals
- regional events and support
- carer support events
- resources including fact-sheets, information booklets, quarterly magazine, kids' magazine, short films and more

Useful Contacts:

Please see page 39 for a full list.

Future Directions

Management of CF has traditionally been based on symptom relief. New modulator *(next generation)* **drug therapy that improves or restores the function of CFTR is targeted at specific genetic mutations. This personalised, or precision medicine, has been described as 'theratyping' and aims at identifying which individual mutations respond to particular CFTR modulators. It is hoped that 90% of mutations will be able to be treated with a specific combination of modulators by 2020** *(Clancy et al, 2019).*

Scientists involved in theratyping are testing approved modulators on CF cells grown in the laboratory situation and looking for biomarkers that indicate the drug is working, so they can predict if the new generation combination drug will work in people who have those mutations. This approach enables drug approval to occur more quickly than in the conventional clinical trial approach. Work continues on new modulators, RNA therapy and gene editing in the hope that an effective treatment will be found for all with CF. People with CF still experience a reduced quality of life. Preventing or slowing lung damage and repairing the CF lung are the main goals of research.

It is hoped that 90% of mutations will be able to be treated with a specific combination of modulators by 2020 *(Clancy et al, 2019).*

CF Clinics in Australia

Victoria

Adult Clinic
Adult Cystic Fibrosis Service
The Alfred Hospital
Commercial Road
PRAHRAN VIC 3181

T: (03) 9076 2315 or
(03) 9076 6960
E: cysticfibrosis@alfred.org.au

Children's Clinic
Department of Respiratory Medicine
Royal Children's Hospital
Level 3, West Building
50 Flemington Road
PARKVILLE VIC 3052

T: (03) 9345 5818
E: respiratory.medicine@rch.org.au

Regional clinics are hosted at Albury and Geelong. Contact the clinic for dates.

Mixed Clinic
Cystic Fibrosis Paediatric and Adult Service
Monash Medical Centre
246 Clayton Road
CLAYTON VIC 3168

T: (03) 9594 2915
E: monashcf@monashhealth.org

New South Wales

Adult Clinics
Respiratory Medicine and Sleep Clinic
Royal Prince Alfred Hospital
Level 11, KGV Building
Missenden Road
CAMPERDOWN NSW 2050

T: (02) 9515 8613

Cystic Fibrosis Adult Outpatient Clinic
Westmead Hospital
Hawkesbury Road
WESTMEAD NSW 2145

T: (02) 8890 6544

Respiratory and Sleep Medicine
John Hunter Hospital
Look Out Road
NEW LAMBTON HEIGHTS NSW 2310

T: (02) 4922 3150

Children's Clinics
Cystic Fibrosis Services
The Children's Hospital Westmead
Cnr Hawkesbury Rd & Hainsworth St
WESTMEAD NSW 2145

T: (02) 9845 0719

Cystic Fibrosis Services
The Sydney Children's Hospital
High Street
RANDWICK NSW 2031

T: (02) 9382 1477 or
(02) 9382 1521

Respiratory and Sleep Medicine
John Hunter Children's Hospital
Look Out Road
NEW LAMBTON HEIGHTS
NSW 2310

T: (02) 4921 3750

Mixed Clinic
Cystic Fibrosis Outpatient Clinic
Gosford District Hospital
Holden Street
GOSFORD NSW 2250

T: (02) 4320 3482

Western Australia

Adult Clinics
Adult Cystic Fibrosis Centre
Sir Charles Gairdner Hospital
Verdun Street
NEDLANDS WA 6009

T: (08) 6457 1756

Children's Clinic
Cystic Fibrosis – Respiratory
and Sleep
Perth Children's Hospital
15 Hospital Avenue
NEDLANDS WA 6009

T: (08) 6456 0217
E: PCH.CF@health.wa.gov.au

Australian Capital Territory

Adult Clinic
Department of Respiratory
and Sleep Medicine
Canberra Hospital
Building 1, Level 2
Yamba Dve
GARRAN ACT 2606

T: (02) 5124 2066
E: RespSleep@act.gov.au

Children's Clinic
Paediatric Care
Canberra Hospital
Building 11, Level 1
Yamba Drive
GARRAN ACT 2605

T: (02) 5124 2222

South Australia

Adult Clinic
Chest Clinic Services
Royal Adelaide Hospital
275 North Terrace
ADELAIDE SA 5000

T: (08) 7117 2900

Children's Clinic
The Breathing Space
Women's and Children's Hospital
King William Road
NORTH ADELAIDE SA 5006

T: (08) 8161 7234

Queensland

Adult Clinics
Adult Cystic Fibrosis Unit
Mater Health
Level 9, Salmon Building
Raymond Terrace
SOUTH BRISBANE QLD 4101

T: (07) 3163 1205

Adult Cystic Fibrosis Centre
The Prince Charles Hospital
Jacaranda Dve
CHERMSIDE QLD 4032

T: (07) 3139 4770

Thoracic Medicine Hospital
Gold Coast Health
1 Hospital Boulevarde
SOUTHPORT QLD 4215

T: 1300 744 284

Children's Clinic
Queensland Children's Hospital
501 Stanley Street
SOUTH BRISBANE QLD 4006

T: (07) 3068 2303
E: lcch_CF@health.qld.gov.au
W: .childrens.health.qld.gov.au/qch/

Tasmania

Adult Clinic
Tasmanian Adult Cystic Fibrosis Unit
Tasmanian Health Service

T: (03) 6166 8308
ask for the CF coordinator

All clinic appointments are centralised

Children's Clinic
Paediatric Clinic
Tasmanian Health Service

T: (03) 6166 8308
ask for the CF coordinator

All clinic appointments are centralised

Care in the Community Contacts

Cystic Fibrosis Community Care (VIC)

T: (03) 9686 1811
E: support@cfcc.org.au
W: www.cfcc.org.au

Cystic Fibrosis Community Care (NSW)

T: (02) 8732 5700
E: nswsupport1@cfcc.org.au
W: www.cfcc.org.au

Cystic Fibrosis WA

T: (08) 6457 7333
E: physio@cfwa.org.au
W: www.cfwa.org.au

Cystic Fibrosis SA

T: (08) 8221 5595
E: cfsa@cfsa.org.au
W: www.cfsa.org.au

Cystic Fibrosis Queensland and Northern Territory

T: (07) 3359 8000
E: admin@cfqld.org.au
W: www.cfqld.org.au

Cystic Fibrosis ACT

T: 0437 485 454
E: support@cfact.org.au

Cystic Fibrosis Tasmania

T: (03) 6234 6085
E: general@cftas.org.au

Notes

References

Boyle, M & De Boek, K. *(2013).* A New Era in the Treatment of Cystic Fibrosis: Correction of the Underlying CFTR Defect. The Lancet Respiratory Medicine 1 *(2)*:158-163

Button, B. M., Wilson, C., Dentice, R., Cox, N. S., Middleton, A., Tannenbaum, E., Bishop, J., Cobb, R., Burton, K., Wood, M., Moran, F., Black, R., Bowen, S., Day, R., Depiazzi, J., Doiron, K., Doumit, M., Dwyer, T., Elliot, A., Fuller, L., Hall, K., Hutchins, M., Kerr, M., Lee, A. L., Mans, C., O'Connor, L., Steward, R., Potter, A., Rasekaba, T., Scoones, R., Tarrant, B., Ward, N., West, S., White, D.,Wilson, L., Wood, J., and Holland, A. E. (2016). Physiotherapy for cystic fibrosis in Australia and New Zealand: A clinical practice guideline. Respirology, 21: 656–667.

Clancy, J., Cotton, C., Donaldson, S., Solomon, G., VanDevanter, D., Boyle, M., Gentzsch M., Nick, J., Illek, B., Wallenburg, J., Sorscher, E., Amaral, M., Beekman, J., Naren, A., Bridges, R., Thomas, P., Cutting, G., Rowe, S., Durmowicz, A., Mense, M., Boeck, K., Skach, W., Penland, C., Joseloff, E., Bihler, H., Mahoney, J., Borowitz, D & Tuggle, K. Journal of cystic fibrosis 18, 22-34.

Keogh, R., Szczesniak, R., Taylor-Robinson, D & Bilton, D. *(2018)*. Up-to-date and projected estimates of survival for people with cystic fibrosis using baseline characteristics: A longitudinal study using UK patient registry data. 17*(2),* 218-227.

Keogh, A., Williams, T., & Pettersson, R. *(2018).* Australian and New Zealand Cardiothoracic Organ Transplant Registry. 23rd Annual Report. Darlinghurst, NSW: ANZCOTR

Lau, E., Barnes, D., Moriarty, C., Dentice, R., Civitico, J., Avedello, A., & Torzillo, P. *(2011).* Pregnancy outcomes in the current era of cystic fibrosis care: A 15-year experience. Australian and New Zealand Journal of Obsterics and Gynaecology, 51, 220-224.

Ornstein, D., Rosentstein, B., & Stern, R. *(2000).* Cystic Fibrosis: Medical Care. Philadelphia: Lippincott, Williams and Wilkins.

Quittner, A., Abbot, J., Georgiopoulos, A., Smith, B., Hempstead, S., Marshall, B., Sabadosa, K & Elborn, S. *(2016).* International Committee on Mental Health in Cystic
Fibrosis: Cystic Fibrosis Foundation and European Cystic Fibrosis Society consensus
statements for screening and treating depression and anxiety. Thorax. 2016 Jan; 71*(1)*: 26–34.

Quittner, A. *(2018).* Cystic Fibrosis Mental Health Roadshow. Sourced from https://www.cysticfibrosis.org.au/mhr#Documents

Ronan, N., Elborn, J & Plant, B. *(2017).* Current and emerging comorbidities in CF. Cystic Fibrosis Quarterly Medical Review, 46: 125-138.

Ruseckaite, R., Ahern, S., Ranger, T., Dean, J., Gardam, M., Bell, S & Burke, N. *(2017).* The Australian Cystic Fibrosis Data Registry Annual Report. Monash University, Department of Epidemiology and Preventive Medicine, June 2018, Report No 19.

Saxby N., Painter C., Kench A., King S., Crowder T., van der Haak N. and the Australian
and New Zealand Cystic Fibrosis Nutrition Guideline Authorship Group *(2017).* Nutrition Guidelines for Cystic Fibrosis in Australia and New Zealand, ed. Scott C. Bell, Thoracic Society of Australia and New Zealand, Sydney.

Sly, P., Gabgell, C., Chen, L., Ware, R., Ranganathan, S., Scott, L., & Murray, C. a. *(2013).* Risk Factors for Bronchiectasis in Children with Cystic Fibrosis. The New England Journal of Medicine, 368, 1963-1970.

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Support for Health Professionals:

We provide education to health professionals across Australia. If you are interested in learning more about cystic fibrosis, contact your local state organisation who can provide options such as content, onsite education or video conferencing.

If you would like more information about CF, go to: **www.cfstrong.org.au/health-professionals/** or **www.cfwa.org.au/health-professionals.**

For NSW & VIC enquiries contact:

Cystic Fibrosis Community Care
282 Neerim Rd
Carnegie, VIC 3163

T: (03) 9686 1811
W: cfcc.org.au
E: admin@cfcc.org.au

For all states refer to **Page 39** in the booklet

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