CYSTIC FIBROSIS Western Australia

Cystic Fibrosis: 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 1 in every 2,500 babies born in Western Australia** *(WA)***. Previously a disease of childhood, however, with advances in clinical care, currently there are more adults with CF than children. In 2015, adults represented 52% of the CF population registered in WA** *(Ruseckaite, Ahern, Ranger, Dean, Gardam, Bell & Burke, 2017).*

Currently there is no cure, however, 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 children born in 2018 can be expected to live beyond 50 years *(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 patients still die of respiratory failure. Slowing the progression of lung disease is the primary aim of CF therapy.

Additional CF treatment goals include:

- Reducing early bacterial colonisation
- Slowing airway inflammation to preserve lung function
- Avoiding pulmonary exacerbations
- Optimal nutrition and management of metabolic complications
- Psychosocial support
- Transplantation and appropriate management of end of life care

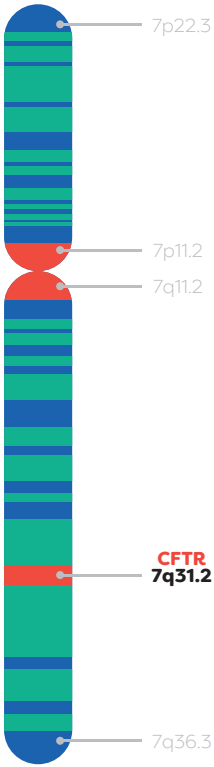
The CF Gene

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 then 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.

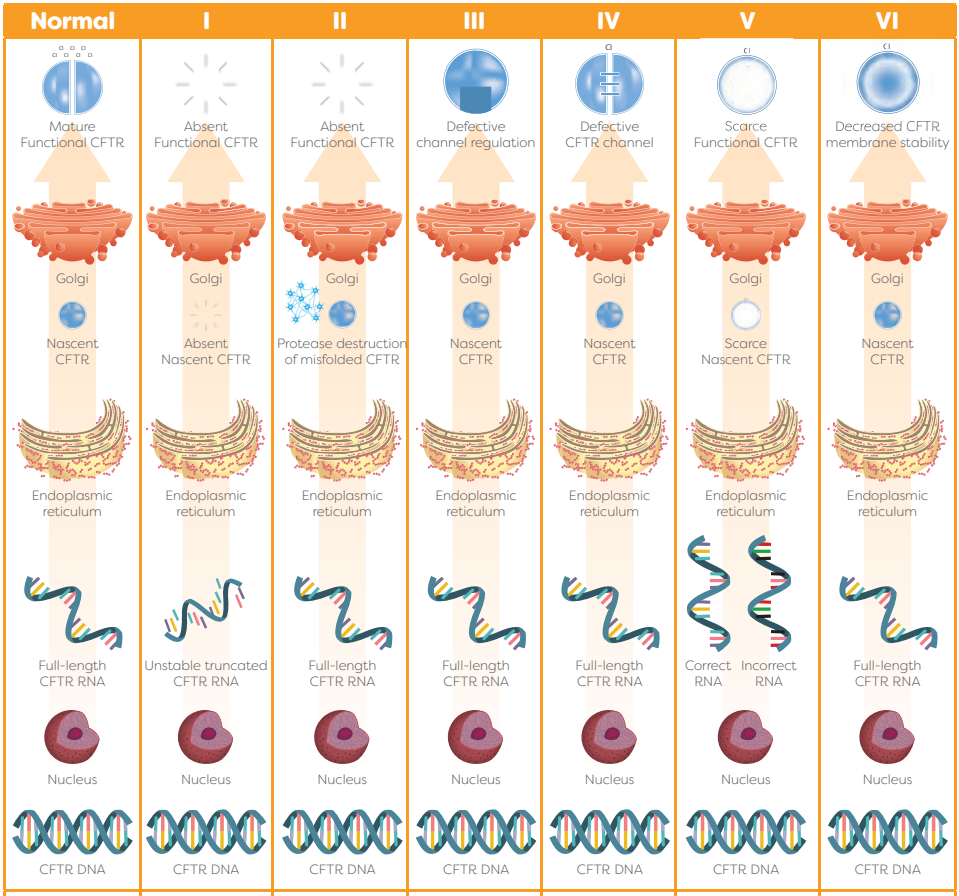
Understanding CF (Vertex): https://www.vrtx.com/cystic-fibrosis/understanding-cf/

> *CF is most common amongst the caucasian races*



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	GlySSlAsp Gly178Arg Gly551Ser Ser549Asn	Arg117His Arg347Pro Arg117Cys Arg334Trp	3849+10kbC▫T 2789+SG▫A 3120+1G▫A 5T	4326delTC Gln1412X 4279insA

(Boyle & De Boek, 2013)

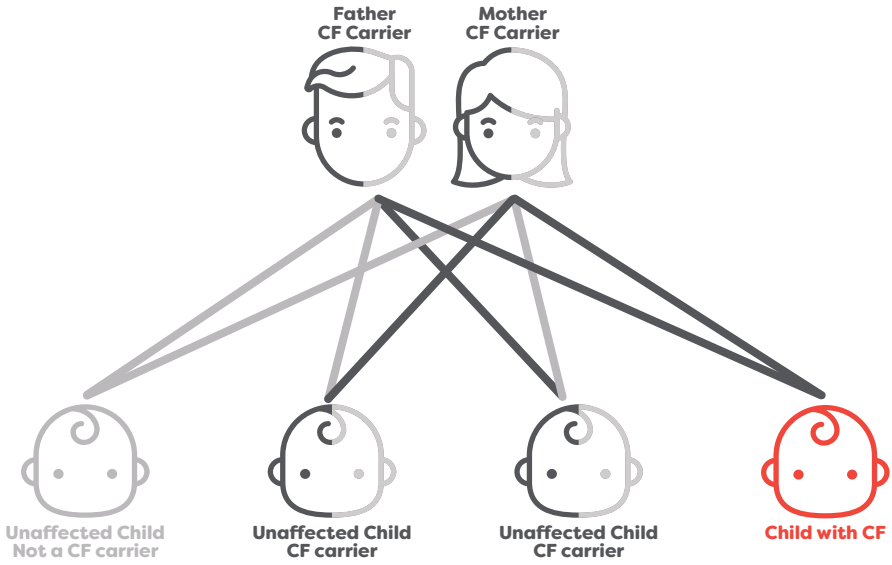
Newborn Screening

CF newborn screening *(NBS)* has been carried out in WA since 2000 and approximately 10-15 babies are born with CF each year. The WA Newborn Screening Program screens for CF along with phenylketonuria, galactosaemia, congenital hypothyroidism, and a range of disorders of amino, organic and fatty acid metabolism.

CF is a common inherited disorder with birth prevalence in Australia reported to be approximately 1:3700 *(Ruseckaite et al, 2016)*. Approximately 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. The most commonly known mutation in CF is delta F508.

Newborn screening for CF is a two-step process. 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. If only one CF gene is identified, they will be a carrier.

WA Newborn Screening Program www.healthywa.wa.gov.au/Articles/U_Z/Your-newborn-babys-screening-test



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

Referral Guidelines for clinicians: https://ww2.health.wa.gov.au/Articles/F_I/Genetic-Services-of-WA-referral-guidelines-for-clinicians

Useful Contact:

Genetic Services of WA
Obstetrics and General Genetics

Ph: (08) 6458 1525
E: gswa@health.wa.gov.au
W: ww2.health.wa.gov.au/Articles/F_I/Genetic-Services-of-WA

10-15 babies
are born with CF each year in Western Australia. *(Approximately)*

1 Person
in 25 carries the gene for CF.

"The WA Newborn Screening Program screens 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 a multi-organ disease, it is the pulmonary disease that presents the most challenge and continues to be the main cause of mortality. Most individuals 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

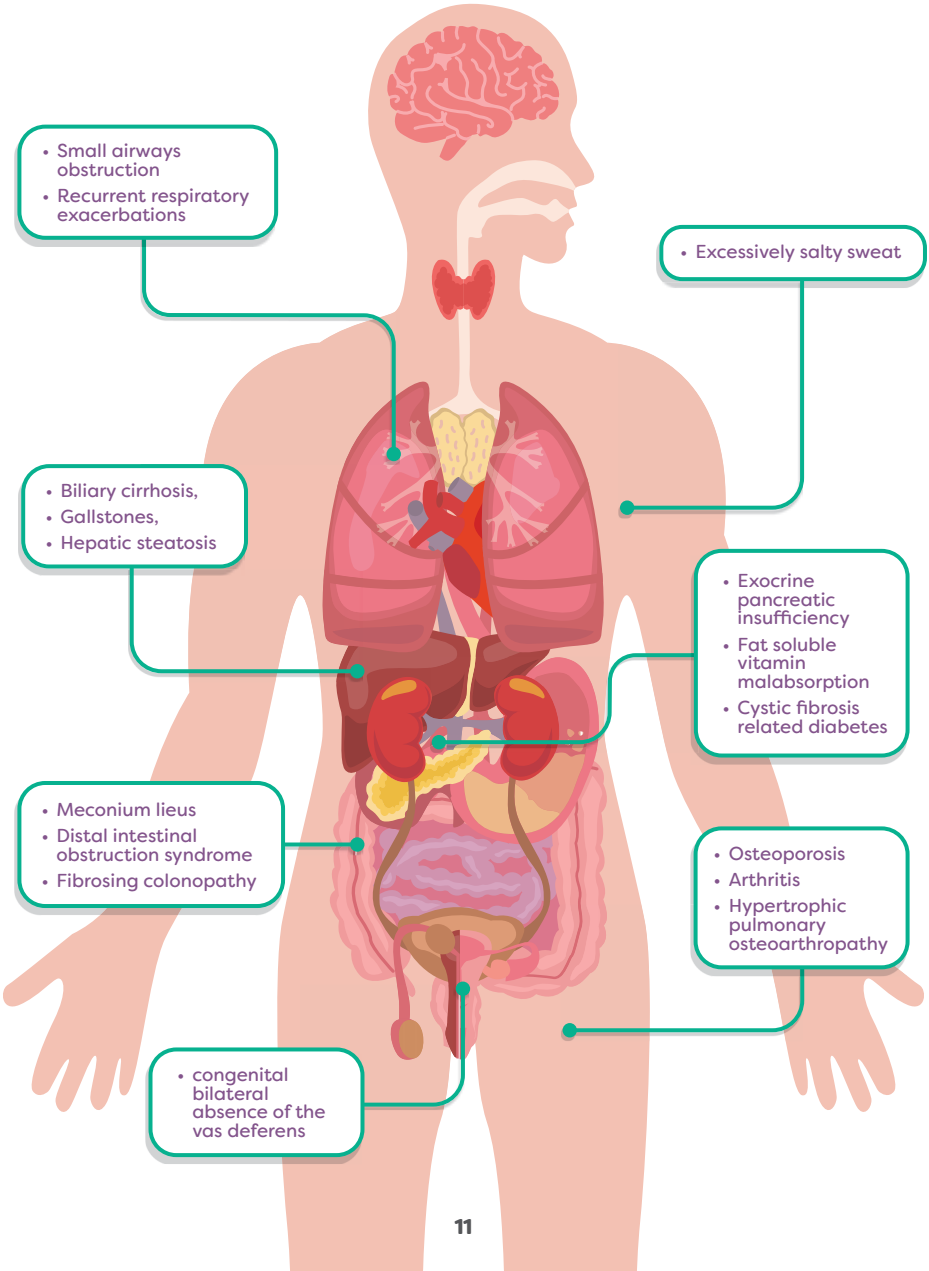
- Pancreatic insufficiency
- Loss of appetite/weight loss
- Abdominal pain
- Gastro-oesophageal reflux disease *(GORD)*
- Pancreatitis
- CF related diabetes *(CFRD)*
- Rectal prolapse
- Distal intestinal obstruction syndrome *(DIOS)*
- Cirrhosis and other hepatic dysfunction
- Cholelithiasis
- Dehydration

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

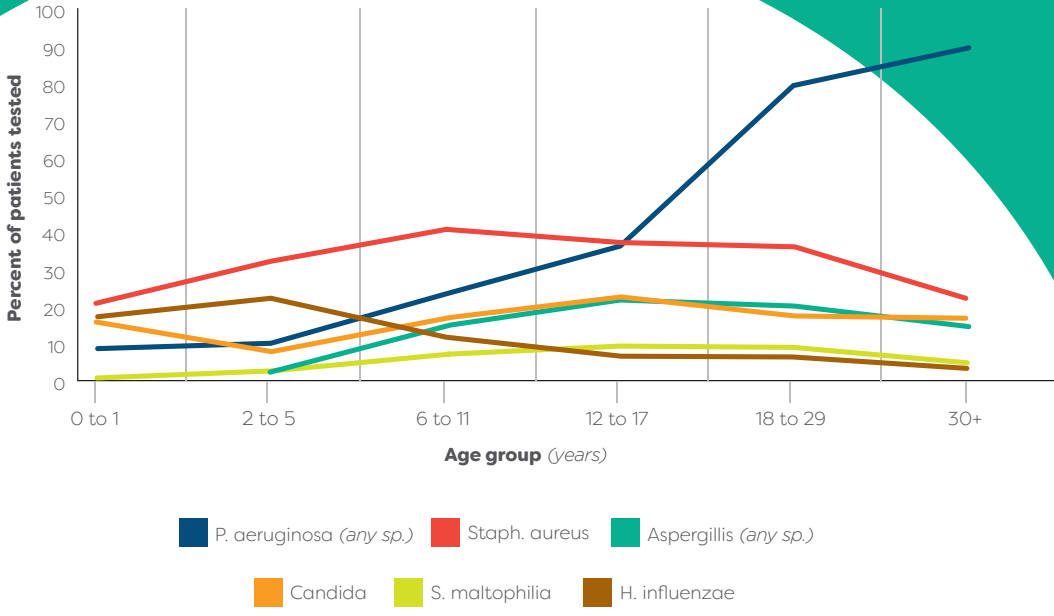


Respiratory

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 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, Gabgell, Chen, Ware, Ranganathan, Scott, & Murray, 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 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.

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 all patients should 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®)*

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.

Hydrator Therapy

Hydrator therapies are used to restore airway surface liquid hydration by osmotically drawing liquid into airways. Bronchitol® and Hypertonic Saline *(HTS)* are commonly used in CF. Most patients 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. Commonly used antibiotics may include amoxycillin and clavulanic acid *(Augmentin Duo®),* flucloxacillin, ciprofloxacin and tobramycin. These and other antibiotics can be administered orally, by inhalation or intravenously.

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.

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.

Currently in Australia the CF community have access to Trikafta®, Kalydeco®, Orkambi® and Symdeko® with many new modulator therapies emerging.

Cystic Fibrosis Drug Development Pipeline https://www.cysticfibrosis.org.au/what-we-do/drug-pipeline

Respiratory Complications

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

Pneumothorax

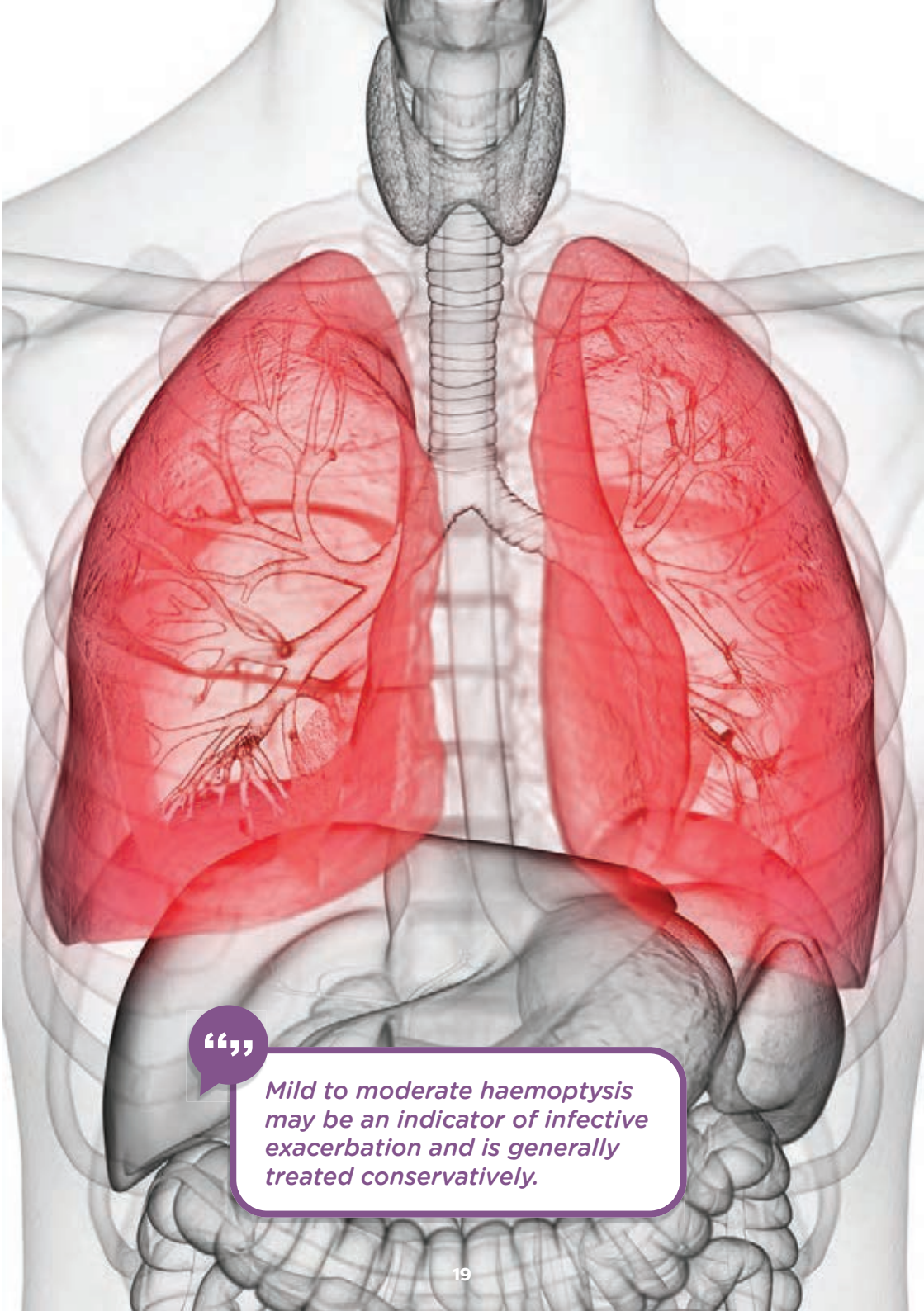
Pneumothorax is defined as the presence of air or gas in the space between the visceral and parietal pleura of the lung, which can impair oxygenation and/or ventilation. A sudden onset of breathlessness in patients with CF should be investigated promptly to identify the possibility of a pneumothorax. 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

Gastrointestinal tract manifestations of CF are related to mucus, dehydration, viscosity and dysmotility.

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-esophageal 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, Painter, Kench, King, Crowder, van der Haak, 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 CF patient is an increasing phenomenon. It is important patients 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 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

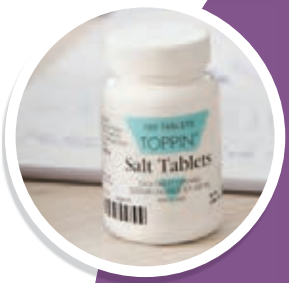
The CFTR defect results in abolition of normal chloride conductance with consequently poor sodium reabsorption into the cells and a high concentration of salt in sweat. This increased loss of sodium and chloride with exercise and in hot temperatures, predisposes the individual with CF to salt depletion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

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 adolescent patients 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 CF patients should be screened for diabetes annually from 10 years of age.

Psychosocial

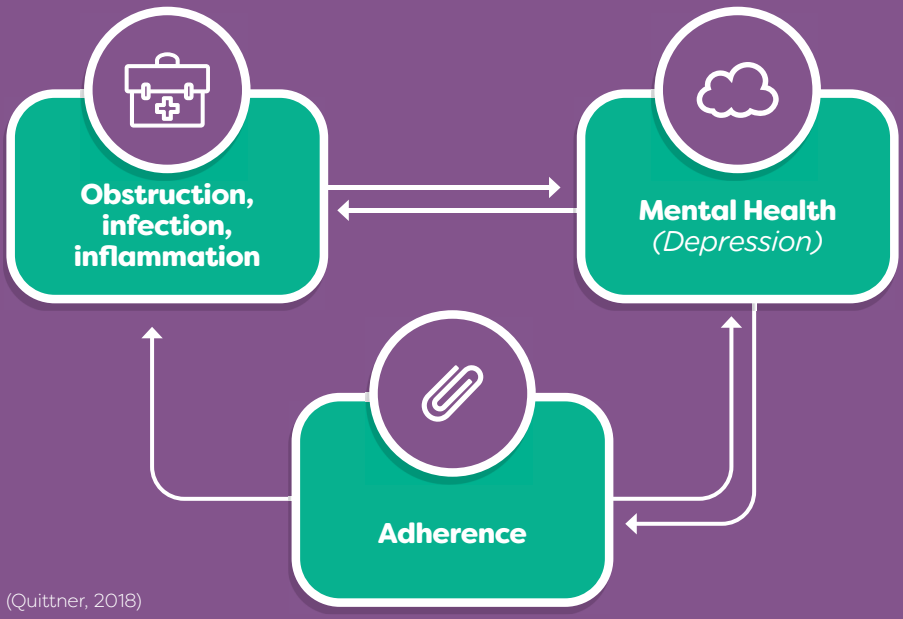
CF is a chronic life-limiting disease, with a complex and high treatment burden. Anxiety and depression are reported to be two to three times higher for individuals with CF and their carers *(Quittner, Abbot, Georgiopoulos, Smith, Hempstead, Marshall, Sabadosa & Elborn, 2016).* **This can be exacerbated at times of diagnosis, hospital admission, specific medical interventions or another diagnosis e.g. 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).*

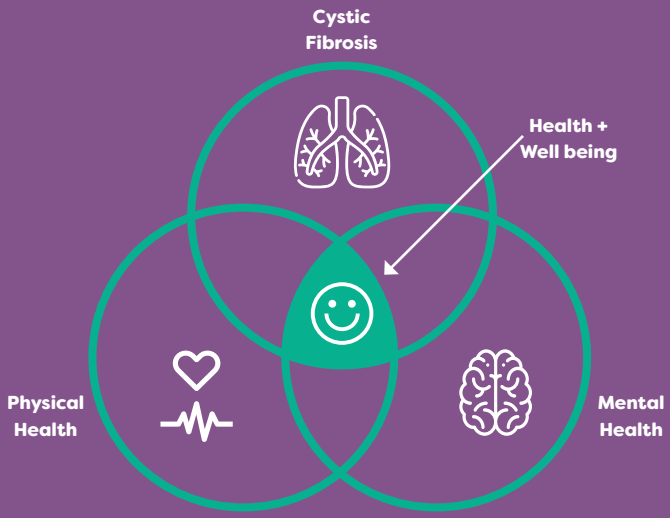
Living with CF can be emotionally and physically challenging for the patient and family. It is important to recognise and treat anxiety and depressive symptoms in people with CF and their carers, as this can impact on treatment adherence, family functioning, quality of life and has been associated with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

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 distance of at least **4 metres apart** at all times to minimise the risk of transmission of micro-organisms. It is important to try to reduce exposure to common respiratory pathogens. Hand hygiene, cough and sneeze etiquette and isolation when unwell remain the most important infection prevention and control strategies. 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 e.g. Nocospray/OxiverTb solution
- Patient should wear a mask in common areas *(ED, outpatients, pathology, radiology)*

Extra precautions for patients with CF known to be infected/colonised with Mycobacterium abcessus 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 room

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

Lung Transplantation

For some individuals 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 is the only paediatric service 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:

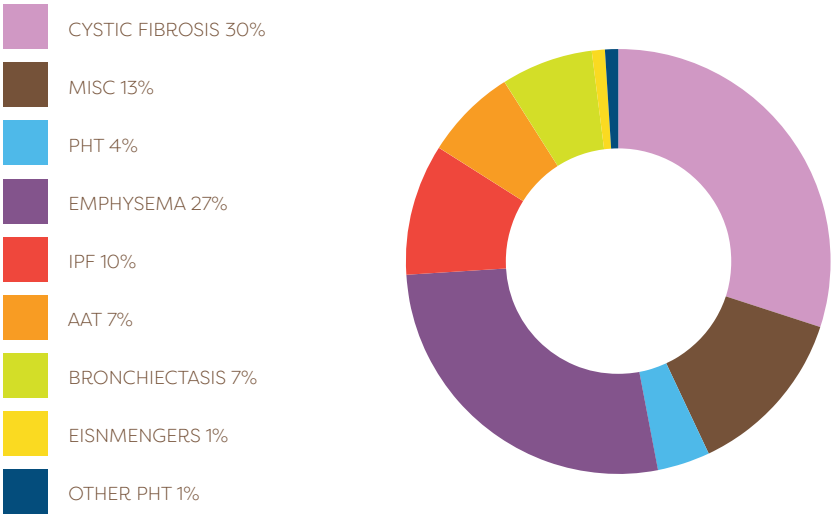
- FEV1 falling to 30% predicted or rapidly falling, particularly in females
- 6-minute walk test <400m
- Development of pulmonary hypertension
- Clinical decline presented by:
  - Long term non-invasive ventilation use
  - Increase antibiotic resistance
  - Pneumothorax
  - Haemoptysis

Useful Contact:

Fiona Stanley Hospital- WA Lung Transplant Program

Ph: (08) 6152 4029
W: www.fsh.health.wa.gov.au

Reason for Bilateral Lung Transplant, 1992 - 2018



(Keogh, Williams & Pettersson, 2018)

51
patients

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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, Barnes, Moriarty, Dentice, Civitico, Avedello & Torzillo *(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.

Useful Contacts:

Genetic Services WA

Ph: (08) 6458 1525
E: gswa@health.wa.gov.au
W: ww2.health.wa.gov.au/Articles/F_I/Genetic-Services-of-WA

King Edward Memorial Hospital- Information for Health Professionals

W: www.kemh.health.wa.gov.au/For-Health-Professionals/Referring/Genetics

Cystic Fibrosis Community Care - Carrier Screening Program

W: www.cfcc.org.au/page/129/genetic-testing

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, patients undergo intensive regimes of treatment and predicting prognosis is difficult. Lung transplant is not an option for all individuals 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, dyspnoea, pain, congestion and anxiety being the most prevalent symptoms. Ideally this would be provided by a specialised palliative care team. In 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 in 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 WA
Ph: 1300 551 704
W: www.palliativecarewa.asn.au

Advance Care Planning
Ph: 1300 208 582
W: www.advancecareplanning.org.au

Continuing Care in the Community

Cystic Fibrosis WA are funded by the Department of Health to provide in home and community-based support for families and individuals with CF. This includes:

- Assistance with airway clearance
- Assistance in developing early routines
- Exercise programs
- Transition support for young people and carers
- Counselling/general support
- Financial support
- Education for daycares, schools, workplaces and health professionals
- Regional events and support
- Carer support events
- Resources including fact-sheets, information booklets,, kids' magazine, short films and more

Useful Contact:

Cystic Fibrosis WA

Ph: (08) 6224 4100
E: servicesmanager@cfwa.org.au
W: www.cfwa.org.au

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 all mutations will be able to be treated with modulator therapy in the future.**

Scientists involved in theratyping are testing approved modulators on CF cells grown in the laboratory situation looking for biomarkers that indicate the drug is working, and thus predict if the new generation combination drug will work in individuals that 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, phage therapy and gene editing in the hope that an effective treatment will be found for all with CF. Individuals 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 all mutations will be able to be treated with modulator therapy in the future

CF Centres in Western Australia

For all CF clinical information and information related to individual clinical care, please contact the relevant CF specialist service.

Sir Charles Gairdner Hospital *(SCGH)*

Sir Charles Gairdner Hospital is the adult CF centre for WA. It provides both inpatient and outpatient services. Outpatient clinics are run twice per week. The adult CF service is run by a Medical Consultant who specialises in CF care, and it is co-ordinated by the Nurse Practitioner.

Contact with this service can be made through the Department of Respiratory Medicine.

Ph: (08) 6457 1756
W: www.scgh.health.wa.gov.au

Perth Children's Hospital *(PCH)*

The CF team at Perth Children's Hospital *(PCH)* is a multidisciplinary team that provides holistic health care to all infants, children and young people in WA with CF until care is transferred to adult services in late adolescence. The team consists of respiratory physicians, specialist nurses, physiotherapists, dieticians, social workers, psychologists, pharmacists, school teachers and administrative assistants as well as affiliated research teams.

They provide health care to children with CF by working in partnership with people with CF and their families from the time of diagnosis. They do so through regular outpatient clinics and in-hospital care when required. They also provide quarterly rural outreach clinics to Bunbury, Kalgoorlie, Port Hedland, Karratha and Broome.

Contact with this service can be made through the Respiratory Department.

Ph: (08) 6456 0217
W: www.pch.health.wa.gov.au

Fiona Stanley Hospital *(FSH)*

The WA Lung Transplant Program is managed through the Advanced Lung Disease Unit at Fiona Stanley Hospital. The program takes referrals from across the State. It provides both inpatient and outpatient services.

The service is co-ordinated by the Clinical Nurse Consultant, Advanced Lung Disease and Lung Transplant Unit.

Ph: (08) 6152 4029
W: www.fsh.health.wa.gov.au

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CYSTIC FIBROSIS Western Australia

CFWA Support for Health Professionals:

We are able to provide education to health professionals in both metropolitan and regional WA, including GPs, nurses, doctors, physiotherapists and other community health workers.

For more information contact us on the details below.

If you would like more information about CF, go to our Cystic Fibrosis for Health Professionals website: www.cfwa.org.au/health-professionals

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