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CHAPTER 1

WHAT IS CYSTIC FIBROSIS?

Cystic Fibrosis (CF) is the most common, recessive genetic condition in Australia, affecting approximately 1 child in every 2500 born in Western Australia. It results from a mutation in a single gene on chromosome 7 that encodes the Cystic Fibrosis Transmembrane conductance Regulator (CFTR) protein. As there are many mutations that can affect the gene, the function of the CFTR protein varies from complete absence to partial function, which is why there is a wide variation in disease severity.

The gene defect results in thicker more viscous secretions from exocrine glands, particularly in the respiratory tract and the pancreas. This results in recurrent lower respiratory tract infections leading to progressive pulmonary damage with bronchiectasis as a major feature. In the gut, the decreased pancreatic enzymes lead to malabsorption and other complications.

When CF was first recognised as a specific condition in the 1930’s the outlook for babies was very poor. Today with early diagnosis, greater understanding of the condition and improved treatment and management, the current predicted survival is in the mid-thirties and increasing. Many patients live much longer than this and lead full and productive lives.

Symptoms

Most babies with CF have no symptoms of CF. Over time they may show some symptoms but these can vary greatly from person to person. The early indicators of CF are often similar to those found in other childhood health problems. Milder forms of the disease may present later, commonly with bronchiectasis of unknown origin in a young adult or male infertility (see later).

Symptoms may include some or all of the following:
- Recurrent wheezing
- Persistent cough and excessive mucus
- Recurrent pneumonia
- Failure to gain weight, often despite a good appetite
- Abnormal bowel movements which sometimes include chronic diarrhoea or
frequent stools which are bulky, greasy and foul smelling - Steatorrhoea.

- Rectal prolapse
- Salty taste to the skin
- Nasal polyps
- Recurrent sinusitis
- Clubbing of the fingertips

The Digestive System

For most people with CF, the exocrine glands in the pancreas produce such thick secretions that the pancreatic ducts become blocked and the enzymes never reach the intestine. This results in malabsorption, particularly of fatty foods and fat soluble vitamins, leading to malnutrition, failure to thrive, poor growth and limited stature if uncorrected.

The liver can be directly affected by CF, although the mechanism is unclear, and can lead to problems such as cholelithiasis and cirrhosis.

Breast Feeding and Cystic Fibrosis

Recent studies have shown that breastfeeding with appropriate enzyme replacements, is the best method for feeding a baby who has CF. Breastfeeding encourages a close bonding between mother and baby. Some mothers have found that breastfeeding has helped them cope emotionally with the sadness and grief that can occur after the baby is diagnosed with CF. It is also important to note that breastfeeding is a very personal and individual choice.

Fertility

Female CF patients will have relatively normal fertility and can carry a normal pregnancy although may be at greater risk if they have significant respiratory disease. Pregnancy should be discussed with their respiratory physician.

Almost all males with CF are infertile due to a congenital absence of the vas deferens, a further complication of CF. However, they will produce normal sperm and can conceive with the help of IVF.
Genetic counselling is essential for patients planning a family.

**Other Important Facts**

- As yet there is no cure for CF, but it is still considered a life shortening condition. With the advances made in recent year’s patients often live long and full lives, with the median survival age in 1999 was 26.1 years in Australia. (Australasian Cystic Fibrosis Data Registry, 1999) The average life expectancy is 32.5yrs in 2005 and increasing.

- It is Important to remember that CF is a genetic condition, and is not contagious. You would be surprised how often this is asked.

- Ongoing damage to the pancreas in CF patients can lead to CF related diabetes. The 2002 CF data registry puts this at 8.1 % of all CF patients. This incidence is higher than the general population. The rate increases with patient age and the incidence in patients over the age of 25 years of age is greater than a 20%. (Australasian Cystic Fibrosis Data Registry, 2002)

The management of CF related diabetes is different to normal diabetes in that there are no restrictions on diet. It is always managed by insulin and not oral hyperglycaemics, usually with variable boluses of short-acting insulin before meals and sometimes a long-acting insulin at night.
CHAPTER 2

DIAGNOSIS

In Western Australia new born screening for CF has been carried out since mid 2000. The protocol for screening is a two stage process. The first step measures immunoreactive trypsinogen (IRT) on the Guthrie blood spot and if this is positive, mutation analysis is performed on the same blood spot.

The level of IRT in blood is increased in newborns with CF. This is thought to represent leakage of the pancreatic enzymes from blocked pancreatic ducts into the bloodstream. However, an increased IRT is not specific for CF. Consequently a second test (that is mutation analysis) is used to identify children with CF amongst the group of patients who have a raised IRT at birth. When two abnormal mutations are detected, the diagnosis of CF is made. When one mutation is detected, the patient is recalled for a sweat test. A normal sweat test would exclude the diagnosis of CF making that person a carrier.

(WA Newborn Screening Program, June 2000)
CHAPTER 3

TREATMENT

CF patients are prone to chest infections associated with excessive viscous mucous production and this can result in chronic lung damage predominantly in the form of cystic bronchiectasis and chronic lung disease. Effective therapy slows down this process and preserves lung function. The fundamental aspect of therapy is airways clearance through physiotherapy and regular exercise. Physiotherapy is recommended as a daily and if possible twice daily procedure and patients (or their parents in childhood) are taught these techniques to perform at home. The addition of nebulised hypertonic saline or nebulised Pulmozyme prior to physiotherapy may help patients whose mucous is particularly difficult to clear. The second aspect of treatment is appropriate antibiotic therapy based on sputum cultures.

The two most common pathogens in CF are Pseudomonas Aeruginosa and Staphylococcus Aureus and protracted illnesses with these organisms may require courses of intravenous antibiotic therapy usually lasting 2 weeks.

Viral infections are the most common cause of chest infections in CF. It is therefore important to try to minimise the risk of catching “colds”. In the home, simple hand washing reduces the risk of transmission. Children with CF must also be immunised according to the normal schedule. Annual flu vaccinations are recommended.

The pancreas is significantly effected in 80% of CF patients and these patients require pancreatic enzyme replacement and dietary supplements including vitamins.

Patients with liver involvement may be treated with ursodeoxycholic acid, which helps symptoms and may slow disease progression. The treatment for CF is as individual as the child, but generally includes some or all of the following:

- Daily chest physiotherapy
- Antibiotics - oral, Inhaled and Intravenous
- Pancreatic enzyme replacement
- Diet and nutritional supplements
- Vitamins
- Other inhaled therapy such as bronchodilators, nebulised hypertonic saline, or Pulmozyme.
- Insulin for patient with CF related diabetes
Specific airway clearance techniques

As the child gets older, they are taught more autonomous airway clearance techniques.

- **Modified Postural Drainage (MPD)**
  For babies up to about 12 months, the basic program involves daily or twice daily modified postural drainage (MPD) with percussion and vibration (P & V), and may or may not include stimulated coughing.

- **Regular Exercise**
  Another very important early recommendation for airway maintenance is regular exercise. Most toddlers are naturally active, others may need prompting. Breathing control exercises may begin in the early years using games and toys that promote useful breathing patterns.

- **Active Cycle Breathing Techniques (ACBT)**
  From about 3-5 years of age, more emphasis is placed on breathing control exercises. The active cycle of breathing techniques (ACBT) which incorporates the forced expiration technique (FET), is taught as a method of clearing mucus more independently. It is most effective when combined with exercise and other physiotherapy techniques.

- **Autogenic Drainage**
  Described by Jean Chevaillier in 1967, autogenic drainage is an airway clearance technique that is widely used throughout Europe. The technique is characterised by breathing control, where the individual adjusts the rate, depth and location of respiration in order to clear the chest of secretions independently. (Agostini P and Knowles N, 2006)

- **PEP Mask or device**
  Devices such as Flutter, Acapella or Pari PEP may be prescribed by the physiotherapist to enhance airway clearance in those people that may benefit from them.

- **Other Techniques**
  Further techniques such as percussion and vibration (P & V) may be increased when a lung infection occurs. Regular exercise is of major benefit and should become a part of life for anyone with CF. Chest physiotherapy may be more effective when combined with inhaled medications.

Because cystic fibrosis affects each person differently, a treatment program is designed specifically for the individual, and needs to be reviewed regularly.

- **Physiotherapy**
  The physiotherapist is an important member of the CF management team, teaching effective clearance of secretions from the lungs and promoting optimal lung health and general physical fitness. 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.
CHAPTER 4

CF SERVICES IN WESTERN AUSTRALIA

Cystic Fibrosis WA

Cystic Fibrosis WA is a not-for profit community support organisation, which is based at The Niche, Nedlands. We provide support to people and families with CF. CFWA also provides an education role in the community, trying to raise public awareness of CF and issues related to the care of a person with CF. As part of a wider group of organisations across Australia, CFWA is involved in lobbying for services, and supporting research.

- Comprehensive Home Care including:

  The CHC program is funded by the West Australian Department of Health. Membership of CFWA is not a requirement for this service. It is designed to provide treatment and/or assistance in the home of people with CF, thereby reducing the incidence of hospitalisation. CHC is usually available by referral from your CF Centre and is described as an early intervention or early discharge package of care. It covers several areas:

  - Assistance with airway clearance;
  - Supervision of Intravenous antibiotics;
  - Newly diagnosed airway clearance service and counselling;

  The services in this program are generally short-term, for example, two (2) weeks, and are only available to patients in the Perth Metropolitan area.

- Counselling (formal & informal), Information, Education & Advocacy Services for:
  - Parents of children newly diagnosed.
  - For clients and their families.
  - On behalf of children & young adults through the educational spectrum (day care to tertiary level)
  - On behalf of people with Cystic Fibrosis in their workplace.
  - On behalf of members with a variety of agencies including but not limited to schools, service clubs, and hospitals.
  - Parent Support Network

- Home Support Service

  Regular visits by a CFWA Home Care Worker to assist with airway clearance.

- Transition Support Service

  Support for clients and their families making the transition from Princess Margaret Hospital to Sir Charles Gairdner Hospital.
Transplant Support
Support for those who are in the Eastern States awaiting lung transplantation. Includes telephone support (1800 No.), help with referral to local services and practical assistance. (Although most transplants are carried out in WA now.)

Supports research.

Advocacy and Lobbying at Local, State, and Commonwealth level on behalf of those affected by CF.

Recreation Programmes
Recreation programmes such as coffee mornings for parents and parents retreats.

Availability of Equipment
A limited range of equipment is available for loan and CFWA will assist members to locate/purchase equipment items.

Sir Charles Gairdner Hospital (SCGH)
Sir Charles Gairdner Hospital is the Adult CF centre for Western Australia. It provides both inpatient and outpatient services. Outpatient clinics are run twice per week on Tuesday and Thursday afternoons. The Adult CF service is run by a Medical Consultant who specialises in CF care, and it is co-ordinated by the Nurse Practitioner who is the Transplant and CF Co-ordinator. Contact with this service can be made through the Department Respiratory Medicine. 08 9346 1756

Princess Margaret Hospital (PMH)
Princess Margaret Hospital for Children is the Paediatric CF centre in Western Australia. It provides both Inpatient and Outpatient services. Outpatient clinics are run on a Wednesday and Thursday afternoon. PMH have five Consultants that look after CF patients, this service is co-ordinated by the Respiratory Clinical Nurse Consultant. Contact with this service can be made through the Department Respiratory Medicine.

Genetic Services of WA (KEMH)
Genetic Services of WA is based at Princess Margaret Hospital for Children; they are responsible for, providing counselling to parents and families of newly diagnosed babies. They have direct links with the Respiratory team at PMH. Genetic Services of WA will also send family members for testing if there is a family history.
CHAPTER 5

CONTACT DETAILS FOR WESTERN AUSTRALIA

❖ Cystic Fibrosis WA (CFWA)

The Niche  
Suite C  
11 Aberdare Road  
NEDLANDS WA 6009

PH: (08) 9346 7333  
FX: (08) 9346 7344  
Freecall: 1800 678 766 (Country WA & NT)  
E-mail: info@cysticfibrosiswa.org  
Website: www.cysticfibrosiswa.org  
Post: Cystic Fibrosis WA  
PO Box 959  
NEDLANDS WA 6909

❖ Sir Charles Gairdner Hospital (SCGH)

Department of Respiratory Medicine  
Respiratory Outpatient Clinic  
PH: (08) 9346 1756  
Fax: (08) 9346 1555

❖ Princess Margaret Hospital (PMH)

Department of Respiratory Medicine  
PH: (08) 9340 8626  
Fax: (08) 9340 8983

❖ Genetic Services of WA (KEMH)

PH: (08) 93408828  
(08) 93408222 (page 3367)  
Fax: (08) 93707058
CHAPTER 6

FURTHER READING

Pamphlets

Published by Cystic Fibrosis Australia
- Cystic Fibrosis Newly Diagnosed 2005
- Cystic Fibrosis Question and Answers 2005
- Understanding Cystic Fibrosis 2005

Published by Cystic Fibrosis WA
- Cystic Fibrosis WA Services Brochure

Books

- Understanding Cystic Fibrosis: A useful guide for families and patients, Karen Hopkins, Ph. D., 1998, University Press of Mississippi
- Cystic Fibrosis: Medical Care, David M. Orenstein, Beryl J. Rosenstein, Robert C. Stern, 2000, Lippincott Williams and Wilkins

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Cystic Fibrosis Australia, 2005

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WA Newborn Screening Program, 2000

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Cystic Fibrosis Australia, 2001

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