An Introduction to Cystic Fibrosis

Introduction

This booklet has been put together by the Monash CF multi-disciplinary team to assist you during the time of diagnosis.

By now you would have met with members of the team (doctor, coordinator, social worker, physiotherapist, and dietitian). The team will have started to discuss CF and its management with your family. People often say they feel tired and overwhelmed during these first few days and often have trouble remembering what has been discussed.

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

Everyone adjusts to new information at their own pace and information may need to be discussed many times before you feel confident and comfortable. Some people find it helpful to write down any questions they have for the team. Feel free to do this as often as you need to. We will be guided by you as to how often you need to discuss the information you have received.

"Parent tip #1: Use a pen and paper, take notes and look over them later. You'll be given lots of materials to read, documents and scripts. Store everything in a folder and maybe keep them together in a plastic tub."

What is Cystic Fibrosis?

Cystic Fibrosis [CF] is a rare genetic illness affecting one in every 2,500 babies born in Australia. It affects both males and females equally. CF primarily affects the lungs and digestive system because of a faulty gene, causing the production of an abnormal amount of excessively thick and sticky secretions in the lungs, airways and digestive system. CF can also affect the liver, sweat glands and the reproductive system. The increase of thick secretions stops the pancreas from working properly and contributes to poor absorption of nutrients, constipation and sometimes bowel obstructions. The excess mucous in the lungs can cause recurrent bacterial infections, inflammation and lung damage. Lifelong medications and physiotherapy techniques will help treat symptoms of CF. A diagnosis of CF involves a high level of commitment from families with the need for regular treatments at home. CF cannot be cured, however with current and new treatments, the outlook has improved significantly over the past few decades and continues to do so.

"Not all organs are affected by the CF mutation gene, the organs it will affect most are lungs, pancreas and sweat glands. It affects boys and girls equally. 1 in every 2500 babies in Australia are born with CF"

It is important to understand that not all organs are affected by CF and these include the heart and the brain. Your child will still be able to attend school and their level of academic achievement should be no different to their non-CF peers. The aims of treatment are to allow people with CF to live a normal life with minimal lung disease and optimal growth.

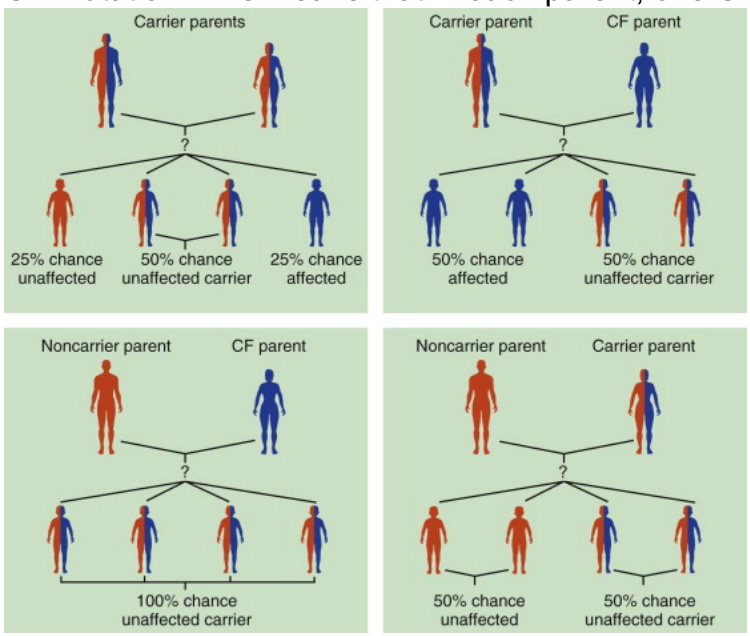
Recently, new medications have been developed to help treat the underlying cause of CF rather than the effects of CF (such as coughing, malabsorption of nutrients, infections, etc). These medications are designed to be used alongside regular CF treatments. In the past few years they have become more widely available to some children with CF.

Genetics of Cystic Fibrosis

CF is an inherited genetic disease. This means that CF is caused by a change or mutation in the CF gene. The gene involved in CF controls the production of a protein called CFTR or **C**ystic **F**ibrosis **T**ransmembrane **C**onductance **R**egulator. This protein controls the movement of salt (sodium chloride) in and out of cells in the body. When a mutation occurs in the CF gene, the function of the CFTR is abnormal, which results in an abnormality in the movement of salt in or out of cells. This abnormality then leads to the problems with sticky mucous and excess secretions as outlined on page 5. There are more than 1500 different mutations in the CF gene, but the most common in Australia is called ΔF_{508}.

HTTPS://CYSTICFIBROSISNEWSTODAY.COM/2015/07/28/NEWLY-DISCOVERED-GENE-THERAPY-HELP-TREAT-

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



Prognosis

CF is a genetic disease which currently has no cure. However, over the past 30 years, advances in treatments for CF have dramatically improved life expectancy, such that the mean age of survival is now over 40 years. There are now more adults than children with CF in Australia.

The improved outlook for children and adults living with CF is due to creating universal best practice guidelines for CF care. With our current management practices, we expect all newly diagnosed CF children will grow and develop to lead a productive and healthy life well into adulthood. The outlook for those living with CF has never been more optimistic and we anticipate further improvements in both quality of life and life expectancy in the future.

For more information visit **www.cysticfibrosis.org.au**
To hear from other families sharing their stories, visit
www.cysticfibrosisbabysteps.org.au

Notes

References

An introduction to Cystic Fibrosis for Parents and Families. (2017).

Cystic Fibrosis Australia. (2017). What is CF? Retrieved from https://www.cysticfibrosis.org.au/about-cf/what-is-cf

Cystic Fibrosis Australia. (n.d). *Cystic Fibrosis New Diagnosis Information for parents.* Retrieved from https://www.cfwa.org.au/wp-content/uploads/2018/03/CFWA-Newly-Diagnosed.pdf

Cystic Fibrosis Baby Steps. (n.d). *What if CF?* Retrieved from http://www.cysticfibrosisbabysteps.org.au/what-is-cf/

Cystic Fibrosis Baby Steps. (n.d).*A Mum's Tips*. Retrieved from http://www.cysticfibrosisbabysteps.org.au/raising-child-cf/a-mums-tips/

Goldfarb, S., Josephson, M.B..(2018). In *BMJ Best Practice Cystic Fibrosis.* Retrived from https://bestpractice.bmj.com/topics/en-us/403/prognosis

Jardens, D. George, G. (2016). *Standard Mendelian Pattern of Inheritance of Cystic Fibrosis* [Figure]. Retrieved from Clinical Key

Monash Health. Monash Children's [Logo]. Retrieved from https://monashhealth.imagegallery.me/

Monash Health. Monash Lung and Sleep [Logo]. Retrieved from https://monashhealth.imagegallery.me/

Royal Children's Hospital. (nd). *Respiratory and Sleep Medicine Parent Information.* Retrieved from https://www.rch.org.au/respmed/parent-information

Vertex Pharmaceuticals Incorporated. (2015). *How to Take Your Kalydeco.*

Photographs by Paige Emmett, (2019)
Image Logo by Melinda Kant. (2019

FAQ

How common is cystic fibrosis?
It is a rare disease with one in every 2500 babies born affected by the disease

What causes cystic fibrosis?
Cystic fibrosis is a genetic condition. One person in 25 carries the faulty CF gene, usually without knowing. If two carriers have a baby, the child has a one-in-four chance of having cystic fibrosis.

Can you catch cystic fibrosis?
No, cystic fibrosis can't be caught or developed; it can only be caused by inheriting two copies of the faulty gene. You could be a gene carrier without having cystic fibrosis.

How will I know if I am a carrier of the gene that causes cystic fibrosis?

Most people do not know if they are a carrier. If you have cystic fibrosis in your extended family, you should ask your GP to be tested as you are more likely to be a carrier than the general population.