

MANUAL FOR

CYSTIC FIBROSIS PATIENTS

AND THEIR PARENTS



WORLD HEALTH ORGANIZATION
HUMAN GENETICS PROGRAMME



INTERNATIONAL CYSTIC FIBROSIS
(MUCOVISCIDOSIS) ASSOCIATION

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MANUAL FOR

CYSTIC FIBROSIS PATIENTS

AND THEIR PARENTS

This Manual is designed to help Cystic Fibrosis patients and their parents to have a better understanding of Cystic Fibrosis as a disease. It has been produced jointly by the World Health Organization (WHO) and the International Cystic Fibrosis (Mucoviscidosis) Association (ICF(M)A), in response to requests for a simple document which is suitable for families in all countries who may have difficulty in attending a well-developed specialist CF clinic or obtaining detailed information about the disease.

If you have any questions about any points mentioned in this booklet, please be sure to discuss these with your own doctor.

This Manual is not intended to replace the information your doctor will give you once you are informed that you or your child has CF but it will supplement and serve as a general guide to have at hand and to be consulted whenever you need it.

In this Manual, we refer to the CF child sometimes as "he", and sometimes as "she", in order to emphasize that both boys and girls can be affected by CF.

Cystic Fibrosis (CF) is a genetic disorder, that is, the child is born with it and does not acquire it later. Both boys and girls can be affected. It causes several symptoms and problems, with a lot of variation in severity between different patients. There is no cure for CF yet. CF cannot be caught like a contagious disease, or passed from one child to another.

Because the child is born with CF, it is also called a congenital disease, and the child who has it has inherited 2 genes for CF, one from the mother and one from the father. Both parents are therefore called "carriers" of the gene, because they only have one copy of the CF gene. A CF child is born only when both parents "carry" a CF gene.

Cystic Fibrosis is a common disease. In many countries approximately one or every 2,500 children born has Cystic Fibrosis. So, if in your area of the world 10,000 children are born each year, four will be born with Cystic Fibrosis.

Cystic Fibrosis affects many of the organs of the body - the lungs, the intestines, the liver and the pancreas produce the most problems. Some children have more trouble with their lungs and some have more trouble with their bowels; each patient is affected differently.

However CF does not affect the intelligence of the child.

What happens in the lungs? The mucus (sputum) that is produced by the lungs of a normal person, is thin, but the mucus produced in the lungs of a CF patient is very thick and sticky. It sticks to the lungs, causing blockage to some of the breathing tubes (airways).

If the mucus stays inside the lungs it will clog the tiny airways and make it easier for micro-organisms (bacterial infections) to grow there. That is why physiotherapy is so important to clear the airways.

Fig. 1. Normal lungs

Fig. 2. CF lungs

The Cystic patient usually coughs a lot to try to remove the sticky mucus.

Fig. 3. CF coughing

What happens to the pancreas? The pancreas is a very important organ for the digestion of food, because it produces many substances called enzymes that help to break down the food we eat. Once the food is broken down the intestines can absorb the digested foods into the body. In persons with CF, the pancreas is also blocked up by thick juices. As a result, digestive enzymes cannot reach the food that we have eaten, and so it is not digested. When food is not digested, it will not be taken into the body, and instead, will leave the intestines in the stools.

So, the pancreas helps to digest the food, which is taken into our bodies to help us grow and remain healthy. In CF the pancreas does not do its job properly.

When children with CF are not properly treated, their stools are large and they have a bad smell of undigested foods; stools contain oil droplets, and float in water; they are difficult to wash away from the diaper, and their colour is often paler than normal stools. The child may have diarrhoea or softened stools and have abdominal swelling and pain,

because of the large quantity of undigested food that is inside the intestines. Sometimes they can become very constipated and may get a blockage of the intestines.

Fig. 4. Pancreas being blocked

The signs of Cystic Fibrosis may first appear at any time in the life of the child, but are usually noticed in the first two years of life:

When to think of CF:

- frequent cough, producing thick mucus
- frequent pneumonia-like illnesses
- no growth, or weight loss, in spite of a normal (or even a large) appetite
- problems with the bowels
- bowel blockage in the newborn.

Another characteristic of children with CF is that their sweat is very salty. The sweat of everybody is salty, but the sweat in CF is much more salty. Sometimes it is first noticed by one of the parents when kissing the child, or salt crystals may be noticed on the skin.

The test that will show if your child has Cystic Fibrosis is called the "sweat test", and it detects excessive salt in the sweat.

Fig. 5. The sweat test

How is Cystic Fibrosis inherited from the parents?

We inherit from our parents and grand parents (ancestors) many things, like the colour of our eyes, and the colour of our hair - which may be blond, brown or black - our height, and many other physical characteristics that we have, and that make each one of us what we are.

Sometimes, unfortunately, we inherit a disease, and this is the case with Cystic Fibrosis.

A child will have CF when he/she inherits two copies of the faulty CF gene - one copy from each of his parents. This sort of inheritance is called "autosomal recessive". In the following figure we show two parents and a CF child:

Fig. 6. CF inheritance

The parents of CF children are normal, although they each have one normal gene and one CF gene. The possibility exists that they may each pass the CF gene to their children. As you can see both parents must have the gene for CF in order for any of their children to have CF. A child will have CF only when he has inherited two CF genes, one from each parent. If a child inherits a CF gene from one parent and a normal gene from the other, the child will not have CF, but will be a "carrier" of CF, like the parents.

In a family where both the father and mother have the gene for CF, one in four of their children may have CF. This is called a chance event. Even if a family already has one or more children with CF, this does not mean that they will have no more children with CF. The chances of inheriting CF are like a game of roulette or of dice; the same number may turn up again and again.

The opposite may also happen. Two parents who are carriers may have many children and, by chance, none of the children may have CF.

Fig. 7. Mother and father with child by the hand walking

To have a child with CF is not anybody's fault. It is painful and it is sad, but there is no one to blame for it. Regardless of the feelings of anger and frustration, you will have to come to terms and learn to live with it. Neither you, nor your child, nor anybody in the family, should feel any shame or guilt about it. Everyone has some abnormal genes, and CF parents were just unlucky because both happened to be carrying a CF gene.

When the diagnosis of Cystic Fibrosis has been made, it is very important that you come to accept that your child has Cystic Fibrosis. Nothing in the world that you do will change this fact. You and your child must learn to live with Cystic Fibrosis.

The sooner you learn to cope with Cystic Fibrosis the less time you will lose in starting a very effective treatment.

Because CF was first discovered not so many years ago, most people, including some doctors, may have heard only a little about it.

Fig. 8. A child with the mother next to him, reading a book

When CF was discovered in the 1930s very little was known about it, and there were very few drugs available for treatment. Now, there is much more that doctors know about CF, and there are many more drugs, especially enzymes and antibiotics, which make patients live a much longer, more normal life.

Fig. 9. A group of children, all of them equal.

When CF is diagnosed, the parents may have been aware for quite some time that their child was unwell, and may have taken him to many doctors. The child may have had many tests. If you are among these parents, you have been through a long and difficult period of life while searching for the cause of your child's problem. It is only natural if this search has made you anxious and distrustful of the doctors, nurses and other health care workers. It is important to remember that CF is not always easy to diagnose.

In some countries CF is not yet thought to be as common as it really is. As a result of improved awareness and education, CF is now known to be present in many populations of the world.

When parents are first told that their child has CF, they usually know very little about the

disease. Sometimes where a relative has CF, the parents will know a little. But as the doctors explain that the disease is chronic and that there is no cure for it, the parents may feel very shocked, sometimes angry; it can take a long time to accept the fact.

Fig. 10. Both parents and the doctor.

Once the diagnosis is made, the sooner you start the treatment, the better.

At first, you may not believe the doctor and think to yourself: Can it be true? Can my child have this disease? Can't she(he) have something else that the doctor has misdiagnosed as CF, and that can be cured?

These thoughts normally come to a parent's mind when the diagnosis is made, or even later. To doubt is human, but one thing is very important: Cystic Fibrosis exists, and if your child has some or all the features and has a positive sweat test, and your doctor has assured you that she has CF, it is important to face the facts. A certain degree of doubt is normal, but you should not let your doubts prevent you from facing the reality.

You may spend a lot of time, money and energy looking for another diagnosis, which would be better spent coming to grips with the real problem.

Treatment of CF

Since Cystic Fibrosis is chronic, that is, a condition that will be with her for the rest of her

life, it needs to be treated properly, in order that the child may survive, and live a life as close to normal as possible.

Usually, Cystic Fibrosis is diagnosed early in life. Sometimes the disease appears to start later in life, or the child's symptoms make the doctor think about another diagnosis. Sometimes, there are no diagnostic facilities available, so there may be a delay in recognizing CF and in beginning the treatment. In other instances, people who are diagnosed later may have a milder form of CF.

In the treatment of CF we must pay attention to the following considerations:

CHEST PHYSIOTHERAPY is used to clear the airways which are full of thick sputum. Physiotherapy (physical therapy) will be prescribed by your doctor, and consists of a series of procedures and/or exercises.

Physiotherapy is to be done regularly throughout the life of the patient.

Physiotherapy is a part of CF treatment which may become very difficult to do, because it requires discipline. In general, it is important to have a physiotherapy session in the morning, as soon as the child wakes up, before breakfast. Another session will occur after he comes home from school, or before going to bed.

The physiotherapy routine will vary according to your doctor's or physiotherapist's advice. The amount of time you spend on it will depend on the state of your child's lungs. It is important that you follow your doctor's or physiotherapist's instructions as closely as possible. If you have not mastered the techniques, you must not be afraid to ask the doctor or the physiotherapist to show you again.

Physiotherapy should be started at the time of diagnosis. It is important that physiotherapy becomes a part of the daily routine for the child and it can be made fun by including games in the treatment regimen.

Children still often find physiotherapy a nuisance and may try different ways of avoiding it. You must be firm from the beginning and not allow your child to command the situation and to manipulate you. This may happen especially when you start the treatment, and when the child may be very frail and weak. You may feel sorry for him and may want to comply with his wishes. Do not give in to your child without a very good reason because it may become a habit. Remember that the treatment is in your child's best interest.

Coughing in CF is a very good thing because it helps to clear the lungs. You should encourage your child to cough from a young age, "to clear out the phlegm". In a child who does not have CF, coughing may be a sign that he is about to get a cold; but in your child, it merely shows that he is clearing his chest.

Never let your child feel ashamed of coughing: encourage him to cough during physiotherapy and throughout the day.

At school or in the presence of strangers, your child may feel embarrassed and want to suppress his cough. The more he tries to stop his cough, the more phlegm is going to be retained, and the more the infection will take over.

SPORTS AND PHYSICAL EXERCISES are very important, because sports tend to help your child to cough and get rid of sputum. Exercise will make the child physically stronger and will help him(her) to breathe better; with exercise, the child's appetite will increase. Talk to your doctor and discuss the possibility of your child taking up some sort of regular exercise as soon as he is able: after the age of five or six, children can do a lot of things that involve exercise. Before that time, only physiotherapy may be possible.

What is the best sport for your child?

It depends on your child and the local possibilities. Any sport is good, provided that your child does it with pleasure.

A sport should not be imposed on a child just because the parent thinks it is good for him.

Especially good are those sports in which the child has to move about, use the arms, and breathe deeply: football, volleyball, swimming, and running are examples.

Fig. 11. Children skipping and playing football

Do not keep your child away from physical exercise: let him decide whether or not he is able; but encourage at least a little exercise. This may help your child a lot.

ANTIBIOTICS are very important medicines to keep CF children alive and well.

Sometimes your child must take antibiotics very frequently, sometimes for a long time. Sometimes they have to be swallowed, but sometimes your child will need stronger antibiotics which will have to be put directly inside the body, that is, inside a vein, and your child then will have to go into a hospital. Also, antibiotics which are inhaled are now being used in some countries.

Antibiotics have helped to prolong the lives of children with Cystic Fibrosis throughout the world, because they kill the bugs that get into the child's chest.

Sometimes parents are worried because they think that so many strong antibiotics will eventually harm their children. This is not so. Many studies have shown that the effects of antibiotics in CF are almost always beneficial. As the germs infecting CF lungs are always trying to outwit the treatment, new antibiotics have to be developed to combat them. For this reason, "ordinary" antibiotics in common use are not the best in CF, and the ones used may sound unfamiliar.

VACCINATIONS are also important for preventing infections. You must arrange to have your child vaccinated, to avoid any of the diseases that might harm her. For instance, if she is not vaccinated she may catch measles, and since she has CF, the disease may be bad for her lungs. Vaccinations will help to keep many nasty diseases away from your child.

Food and Enzymes

ENZYMES (pancreatic supplements) are very important to help the child digest food, gain weight and grow normally. Your doctor will teach you how your child should take the enzymes, but here are some rules:

- The enzymes should be taken BEFORE, and if the doctor advises, DURING meals.

Fig. 12. The enzymes and meals

- The enzymes should NOT be taken after meals.
- The enzymes should not be chewed; if necessary open the capsule and let your child swallow the granules. Teach her early to swallow the whole capsule, which she may be able to do when 4 or 5 years old.
- You should encourage her to learn to swallow enzyme capsules: start playing a game with her, asking her to swallow a cooked rice grain with her favourite drink, a pea or a lentil will do, and then something bigger, maybe the size of a cooked bean. Do not make swallowing a big event, so that the child will think it is a very difficult thing to do. The child should be complimented when she swallows a capsule for the first time. And do not worry; if the child swallows by herself without you forcing her, she will not choke on the capsule. Remember, if you are insecure or afraid, your child will also feel insecure and afraid.
- Never give food to your child, except fruit, fruit juices, and water, without first giving the enzyme. If you forget to give the enzymes in one snack, that snack is not digested, and is not going to be taken into the child's body.

AGAIN

- Always encourage your child to take enzymes regularly before a meal or a snack, for he/she is likely in the future to do what you taught him to do. You are your child's model, and he will copy your behaviour.
- When your child goes to school, he will have to take his enzymes by himself. The teacher should know about his condition, and should be informed about having to take capsules before meals and snacks.

What kind of food shall you give to your child?

There are no special foods for a child with CF. Your child should receive a good normal diet. Food with a lot of protein and fat: milk, beef, chicken, fish, eggs, cheese. Food with a lot of calories: starchy foods like potatoes, pumpkin, pasta, rice, beans, bread and milk. Any type of milk is good, provided it has been boiled or pasteurized.

No foods are forbidden to the CF patient; there are no restrictions; your child can eat everything; butter and fried foods should be encouraged, not restricted.

The food your child eats helps him to grow up and to strengthen his body defenses to fight against infection.

For good nutritional management you should do as your doctor or dietitian tells you. Depending on the age of your child, he is to have the same number of meals as a normal child is allowed. But the caloric intake of your child should be higher; he needs to be stronger to fight infection. Also, nutritious snacks between meals are important, although too many snacks should be discouraged, and you should avoid giving your child sweets between meals, or instead of the meals; a lot of sweets given at all times is not good.

Meals should be given at established times, beginning with the enzymes, and as the child grows older, you should tell him that to eat well is part of his treatment.

Do not allow your child to eat every hour, because that will not do him any good. But do remember that 2 or 3 high-calorie snacks throughout the day are fine.

Do not promise your child a present if he eats his meal.

Your child needs to learn that he is eating for his own good. If you do give him a present

when he eats, he may always want a present to complete his meal. You should compliment him, saying "very good", after he has finished everything on his plate.

Children with CF lose more salt than others, especially in summer when they sweat a lot, or when they have a fever, or after a lot of exercise. Thus your child will need to drink plenty of fluids, and he may also be asked by his doctor to take salt tablets. Artificially flavoured drinks are not recommended: not because they will do harm to your child, but because they will occupy the place of more important and nutritive foods.

As you can see, good nutrition is very important to CF, as it will keep your child well; but for the treatment to be successful, you should follow your doctor's advice very closely.

The treatment of CF is not easy. It is difficult when you are not used to it. When you learn how to do it, you can manage as in a routine. It takes time, but all parents learn how to treat their CF child. The treatment is continuous and forever, as there is still no cure for CF.

The treatment of CF is very expensive when we consider the antibiotics and the hospitalization, and no family could pay for it for a long time without very serious financial problems.

As treatment is expensive, you may need to seek and insist on help from your local government/health authority. If many parents group together and approach a government authority, this group of parents is more likely to be received and heard by an authority, and have their needs answered.

Fig. 13. Group of patients

You must remember that if your child is well now, it is because she has been well treated by the doctor and the treatment: physiotherapy and enzymes should never be stopped, even if the child looks very well.

If your child looks unwell or weak, she must be seen more often by the doctor. If she is getting worse you must contact the doctor and talk with him. Check with your doctor that:

- the treatment is being followed correctly

- the enzymes and antibiotics are being given in the right amounts

- the physiotherapy is being done correctly.

If the treatments are all correct - the physiotherapy, the enzymes, the antibiotics - perhaps your child needs to be checked to see if there is a new bug in her sputum which has not been identified before, or to find out what else is wrong with her. She may have to be admitted to hospital for intravenous antibiotics.

Hospital

The centre where your child is seen should be in or near a hospital, so she can use the hospital when there is the need.

If a child is too unwell to do her physiotherapy or take her enzymes, she must immediately be taken to the hospital.

One of the questions which parents may ask is whether, in a hospital, the child might "catch a worse disease than CF". Admission to hospital usually means that antibiotics will be given, which will help to fight the infection in your child's lungs. If the doctor advises it, you should never fear to let your child go into hospital for treatment.

Sometimes parents are doubtful about the treatment, and they ask themselves:

Can I find a cure somewhere else? Unfortunately, there is no cure for CF, anywhere in the world. Many centres in developed countries are working very hard to find a cure, but no cure has yet been found. There is a good possibility that in future, gene therapy, or other new treatments, may be helpful.

Fig. 14. Doctors with a test tube and microscope

Some parents do not believe in the diagnosis of CF and abandon the prescribed treatment; but stopping the treatment, or leaving the CF treatment centre will only shorten the patient's life.

When dealing with CF we must see what we can do to help our child and not look for something unreal or magic. Religion and prayer do help in comforting the family. When dealing with CF we must also see what we can do to help the child now.

Alternative treatments will not cure the child. Just as with eye colour, CF is something the person is born with. It is in the "genes".

Sometimes the treatment is too time-consuming and you may be very tired; besides you also must care for your home, and the other children.

Relatives, like the grandparents, aunts and godparents may help a lot. They may help

with the child, they may help even with physiotherapy, or play with your child when you are too tired, or have other tasks to do.

You should trust your nearest relatives and friends and tell them what CF is. You may even teach them to help you to "treat" CF; but if they do not know what the disease is about, they may not understand why physiotherapy is done, when the child looks so well, or why he has to go into hospital when he/she looks so healthy. Then you must tell them that your child looks well and healthy because he is doing the treatment correctly in order not to fall sick.

Fig. 15. Doctor, parents and relatives

You will choose whom to tell, and when to tell that your child has CF. Later your child will make the choice of whom to tell. Some people do not like to tell others of their personal problems, and that choice has to be respected; but whenever you have close friends and you trust them, sometimes it is good to confide in them; it is reassuring and comforting to have someone to talk to, especially when your child has a chronic condition.

If you feel that your relatives are not helping with their advice or are trying to prevent you from carrying on with the treatment, or are making you feel insecure, convince them to come to the clinic with you and explain the situation to the doctor, and ask the doctor to have a chat with them.

Do not behave as if your child is sick and helpless. Don't say "let's give him a break", or stop or decrease the physiotherapy or the enzymes, or decide by yourself not to bring him to hospital when needed. You must make yourself strong to help your child to fight CF.

Do not forget that your child happens to have Cystic Fibrosis. But in every other way, he is a normal child, with normal intelligence, and he will be expected to go through all the experiences that a normal child goes. For instance, he will have colds, or a sore throat, or he may have an injured arm or leg after a football game, like any other child, without CF being responsible for it.

As your child grows older, you should encourage his independence as you should also encourage him to be responsible for his treatment. This should happen gradually. Never lay the burden of the treatment entirely on your child's shoulders. He will not be able to do the treatment by himself especially when he is young. When he is 18 or 19 he may be more independent - perhaps a little earlier, perhaps a little later - but do not expect a child or adolescent to behave like an adult. You may try to give him some responsibility, but this has to be supervised responsibility. Your child must accept it and not have it imposed on him, and then you can see whether he is taking his enzymes correctly and doing his physiotherapy as it should be done.

Since CF is not anybody's fault, neither you nor your child, nor anybody in the family should feel any shame about it. If the child feels ashamed, he will try to hide it from his friends and from everybody else; he will stop taking the enzymes, and will not cough in public. You should make your son feel loved as much as any other child. He is special.

Fig. 12