A Manual for Cystic Fibrosis Patients and their Parents

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Foreword

This manual is designed to help give cystic fibrosis patients and their parents a better understanding of the disease. This leaflet is part of a European initiative undertaken by the European Concerted Action for Cystic Fibrosis and has been widely translated and distributed. The text is based on the manual of the World Health Organization (WHO) and the International Cystic Fibrosis (Mucoviscidosis) Association (ICF(M)A).

If you have any questions concerning items in this leaflet, we encourage you to discuss them with your medical doctor. The addresses of patient support groups are provided at the end of the manual.

We are grateful to AstraZeneca Diagnostics for the production and distribution of this manual. We hope it will help answer questions about cystic fibrosis and help clarify the situation for patients and their families.

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www.med.kuleuven.ac.be/cme/cf/cfnetwork.htm

What is cystic fibrosis?

Cystic fibrosis is a common disease, which in most European populations, affects approximately one in every 2500 children born. This means that for every 10,000 children born each year, four will have cystic fibrosis.

Cystic fibrosis (CF) is a genetic disorder that affects both boys and girls. Children are born with CF and is not contracted during life. That is, CF cannot be caught like a contagious disease, or passed from one child to another. Because a child is born with CF, it is also known as a congenital or inherited disease.

Cystic fibrosis is found in children who have inherited two CF genes, one from each parent. Individuals who only have one copy of the CF gene are called “CF carriers” and are perfectly healthy. A CF child is born only when both parents “carry” a CF gene.

There is a wide variation in the symptoms and severity of the disease between different CF patients. Cystic fibrosis affects many of the organs of the body but causes most problems in the lungs, intestines, liver and pancreas. Some children have more trouble with the lungs and some have more trouble with their bowels; each patient is affected differently. CF does not however, affect the intelligence of the child. At present there is no cure for CF.

What happens in the lungs?

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

If mucus stays inside the lungs, it clogs the tiny airways and makes it easier for micro-organisms (bacterial infections or 'bugs') to grow there. This is why it is important to clear the airways through physiotherapy.

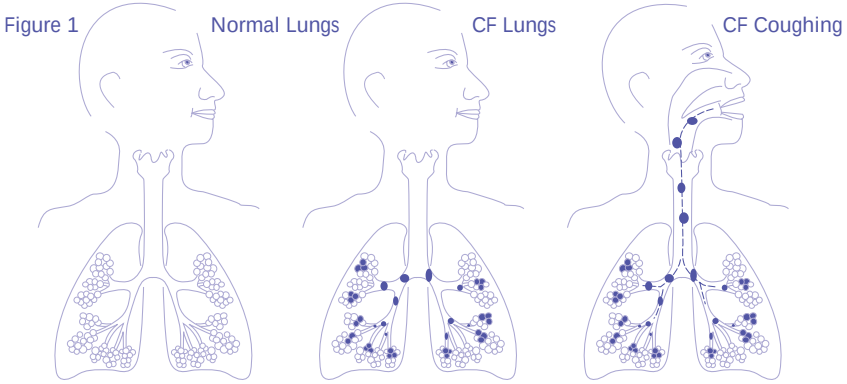


Figure 1

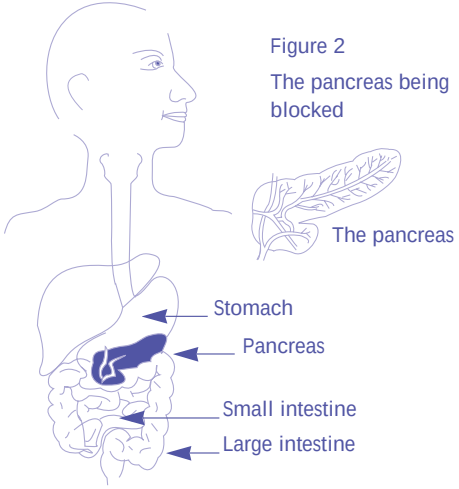


Figure 2

The pancreas being blocked

What happens in the pancreas?

The pancreas is a very important organ for the digestion of food. It produces many substances called enzymes that help to break down the food we eat. Once the food is broken down, the intestine can absorb the digested foods into the body. In people with CF, the pancreas is blocked up with thick juices. As a result, digestive enzymes cannot reach the food eaten and it is not digested. Undigested food cannot be taken into the body and is passed out in stools (faeces).

The pancreas therefore helps to digest the food which is needed to help us grow and remain healthy. In CF, the pancreas is prevented from doing its job properly.

When children with CF are not properly treated, their stools are large and have a bad smell of undigested foods. The stools contain oil droplets, float in water and are difficult to wash away from a diaper (nappy). Their colour is often paler than normal stools. A CF child may have diarrhoea, or softened stools, and have abdominal swelling and pain. This is due to the large quantity of undigested food present in the intestines. If they become very constipated the intestines may also become blocked.

When to suspect cystic fibrosis?

The first signs of cystic fibrosis can appear at any time but are usually noticed in the first two years of a child's life.

You may suspect CF because of the following symptoms:

- Frequent coughing, 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 of a CF child is very salty. It is often first noticed by parents when kissing the child, or from seeing salt crystals on the skin.

The test used to confirm whether a child has cystic fibrosis is called the "sweat test", and it detects excessive salt in the sweat.

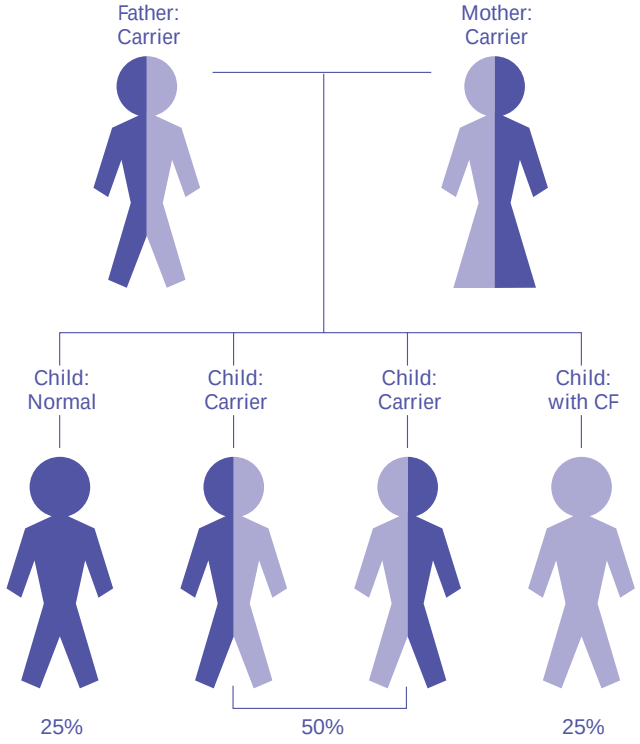
How is cystic fibrosis inherited from the parents?

Everyone inherits the way they look from their parents and grand parents (ancestors). Things like eye and hair color, height and many other physical characteristics that make each one of us what we are.

Sometimes we can also inherit a disease and this is the case with cystic fibrosis. A child will have CF when it inherits two copies of a faulty CF gene - one copy from each parent. This sort of inheritance is called "autosomal recessive".

Figure 3. shows two parents and a CF child :

Figure 3.



The parents of CF children are normal healthy individuals although each carries one normal gene and one CF gene. It is possible therefore, they may both pass the CF gene to their children.

- Both parents must first have a copy of the gene for CF in order for any of their children to have CF
- A child will only have CF when it inherits two CF genes, one from each parent.
- A child will not have CF if it inherits a CF gene from one parent but a normal gene from the other. In this case it will be a “carrier” of CF like its parents.

In European populations, about 1 in 25 individuals are CF carriers. This results in a CF child occuring in approximately 1 in 2500 newborns. The exact frequencies vary between different populations within Europe.

In a family where both the father and mother carry the gene for CF, there is a one in four chance that any child may be born with CF. This is called a ‘chance event’.

- Even if a family already has one or more children with CF, the risk of having another child with CF is the same.

When both parents carry a gene for CF, the chances of their children inheriting CF is like a game of roulette or dice; by chance 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.

There is now a molecular genetic test which can detect the most common disease-causing changes in the CF gene and will identify most of the people who are CF carriers.

The test can be done on a small blood sample, or from a ‘cheek’ sample (obtained by rubbing a small brush on the inside of a person’s cheek).
This genetic test can also be done on prenatal samples (amniotic fluid or chorionic villi). Accurate interpretation of the test depends on knowing that the man tested is the true biological father of the unborn baby.

To have a child with CF... and to accept a child with CF

To have a child with CF is nobody’s fault.
It is painful and sad, but no-one is to blame.

Neither you, your child, or family, should feel any guilt or shame about the fact. It is quite common to have feelings of anger and frustration which are difficult to live and learn to come to terms with. Everyone has some abnormal genes and CF parents were just unlucky because they both happened to be carrying a CF gene.

When the diagnosis of cystic fibrosis has been made, it is very important you come to accept that your child has cystic fibrosis. Nothing in the world you can do will change this fact. The sooner you and your child learn to live with cystic fibrosis, the sooner you'll learn to cope and the less time will be lost in starting an effective treatment.

Because CF has only recently been discovered, most people, including some doctors, may have heard only a little about it.

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

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

When parents are first told that their child has CF, they usually know very little about the disease. As doctors explain that the disease is chronic and that there is no cure, they may feel very shocked and sometimes angry. It can take a long time to accept the fact.

At first you may not believe the doctor and think to yourself:

- Can it be true?
- Can my child have this disease?
- Can't he/she 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:

If your child has some or all the features, has a positive sweat test, and your doctor has assured you it is CF, it is important to face these facts.

A certain degree of doubt is normal, but you should not let your doubts prevent you from facing the reality. Nowadays it is possible in most cases to confirm the diagnosis with a genetic test.

You could spend a lot of time, money and energy looking for another diagnosis. This will cause delay and would be better spent helping your child start their treatment programme as soon as possible.

Once the diagnosis is made, the sooner treatment is started the better.

Treatment of cystic fibrosis

Cystic fibrosis is a chronic condition that the child will have for the rest of their life. It needs to be treated properly in order that the child may survive and live a life as close as possible to normality.

Treatment of CF requires the following therapies:

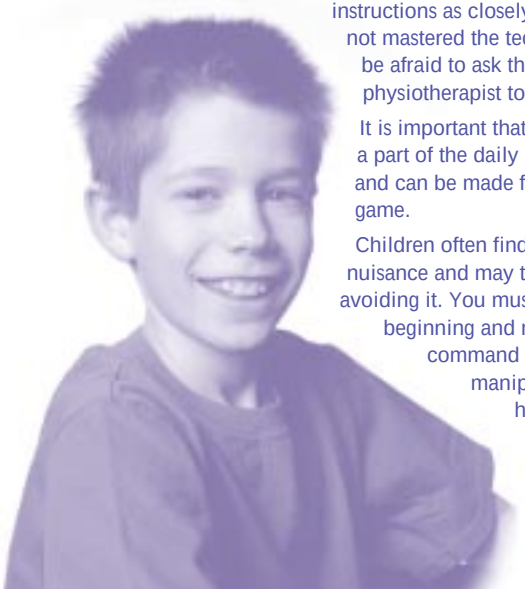
CHEST PHYSIOTHERAPY — Physiotherapy has to be done regularly throughout the life of the patient

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

In general, it is important to have a physiotherapy session:

- in the morning, as soon as the child wakes up, before breakfast.
- after 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.



It is important that physiotherapy becomes a part of the daily routine for your child and can be made fun by turning it into a game.

Children 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 manipulate you. This may happen when you first start the treatment and your child is very frail and weak. You may feel sorry for your child and want to comply with their wishes. Physiotherapy is difficult because it is demanding on you and your child, and requires discipline. Remember, this treatment is in your child's best interest, so do not give in without a very good reason because it may become a habit.

COUGHING — 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". At school or in the presence of strangers, your child may feel embarrassed and want to suppress their cough. This will only cause more phlegm to be retained increasing the chance of infection. Never let your child feel ashamed of coughing during physiotherapy and throughout the day. Coughing in a normal child may be a sign that they are about to get a cold, but in your child it is essential to clear the chest.

SPORTS — Sports and physical exercises are very important

To start with only physiotherapy may be possible. However, if your child is willing and able, sports will help your child to cough and get rid of sputum. With regular exercise the child becomes physiologycally stronger and will be able to breath better. It is good to encourage at least a little exercise but do not impose it on them. Never prevent your child from taking physical exercise as this may help them greatly.

What is the best sport for your child?
This depends on your child and the local amenities. Any sport is good, provided that your child enjoys it. Active sports in which the child has to move around, use the arms and breathe deeply are recommended. Football, volleyball, swimming, and running are good examples.

ANTIBIOTICS — Antibiotics are very important medicines to keep CF children alive & well

Antibiotics kill the bugs that get into a child's chest and have helped to prolong the lives of children with cystic fibrosis throughout the world.

Sometimes your child will have to take antibiotics frequently and for long periods. Often they can be swallowed, but if stronger antibiotics are required they will have to be put directly into the bloodstream through a vein (intravenously). In this case, the child will have to go into the hospital. Some countries are also beginning to use antibiotics which are inhaled.

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.

Germs infecting CF lungs are always trying to outwit old antibiotics and 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 —
Vaccinations are also important to prevent infections

Vaccinations will help to keep many dangerous diseases away from your child. Your child must be vaccinated against measles as this disease is particularly bad for the lungs of a CF child.

FOOD AND ENZYMES — Enzymes (pancreatic supplements) are very important to help the child digest food

ENZYMES

Pancreatic enzymes will help your child to 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.
- 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.
- You should encourage your child to learn how to swallow the whole enzyme capsule from an early age. This may be started when the child is 4 or 5 years old. Swallowing can be introduced as a game where your child is encouraged to swallow a cooked rice grain (or a pea or lentil) with their favorite drink. You can then progress to something about the size of a cooked bean.

  Do not make swallowing a big event or the child will think it is a very difficult thing to do. The child should be praised when a capsule is swallowed the first time. If your child swallows naturally it will not choke on the capsule. Remember, if you are insecure or afraid about this, 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 enzyme with a meal or snack, it will not be digested and taken into your child's body.

REMEMBER

- Always encourage your child to take enzymes regularly before a meal or snack. You will need to set an example to your child in remembering this.
- At school, your child will have to take enzymes by itself. Tell the teacher about your child's condition and the need to take the capsules before meals and snacks.

FOOD —
Good nutrition is very important to keep your child well

What kind of food should you give to your child?

There are no special foods for a child with CF. Your child should receive a normal balanced diet. This should include:

- Foods with a lot of protein and fat such as: milk, beef, chicken, fish, eggs, cheese.
- Foods with a lot of calories such as: 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 and your child can eat everything including butter and dried foods. The food your child eats will help it strengthen the body's defenses to fight against infection.

Meals should be given at established times always beginning with the enzymes. Your child should have the same number of meals as a normal child but the calorific intake of each meal will need to be higher to build strength to fight infection. Nutritious snacks between meals are also important although too many snacks taken regularly through the day should be discouraged. 2 or 3 high-calorie snacks throughout the day are fine. You should avoid giving your child sweets between, or instead of meals.

As the child grows older it will need to understand that eating well is all part of the treatment. It is better not to promise your child a present for eating a meal - giving praise after your child finishes a meal is more effective.

Children with CF lose more salt than others, especially when they sweat a lot. This happens in the summer, or after a lot of exercise. Your child will need to drink plenty of fluids and your doctor may also prescribe salt tablets. Artificially flavoured drinks are not recommended because they fill a child up and might take the place of more important and nutrititious foods.

For good nutritional management you should follow your doctor's or dietitian's advise very closely.

THE TREATMENT OF CF IS NOT EASY
but all parents learn how to treat their CF child in time

Although it will be difficult when you first start treatment, it does become routine as you learn and become practiced at it. Treatment will then be needed for the rest of your child's life and this can be very demanding.

Sometimes parents are doubtful about the treatment and wonder if they can find a cure somewhere else? Many centres in developed countries are working very hard to find a cure, but one has yet to be found. There is a good possibility that in the future, gene therapy or other new treatments, may be helpful.

Some parents do not believe in the diagnosis of CF and abandon the prescribed treatment. There are no magic alternatives and stopping treatment, or attendance at the CF treatment centre, will only shorten the patient's life. Just like eye colour, CF is something the person is born with and cannot be changed. It is in the "genes".

You must also remember that if your child is well now, it is because the of the treatment and care it has received. Physiotherapy and enzymes should never be stopped, even if the child looks very well.

You will need to visit the doctor more often if your child looks weak or ill, especially if the condition appears to be getting worse. Check with the 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 correct, your child may need to be checked to see if there is a new lung infection, or to look for other causes. Additional treatment may need admission to hospital for intravenous antibiotics.

If your child is too unwell to do physiotherapy or take enzymes, they must immediately be taken to the hospital.

Hospital

One of the questions parents ask is whether their child might "catch a worse disease than CF" during a hospital stay.

Admission to hospital usually means that special antibiotics are needed to fight the infection in your child's lungs. The risk of not letting your child go to hospital for this treatment far outweighs the extremely rare chance of catching a worse disease.

The cost of treatment

The treatment of CF is very expensive when we consider the cost of antibiotics and hospitalization. In some European countries the cost of treatment is covered by the health service. Where it is not, you may need to seek help from your local government/health authority. A stronger case can usually be made if the approach to the government authority is made by a patient support group rather than individual parents.

Relatives and friends

You will choose who and when to tell that your child has CF. Later your child can decide for itself who to tell. Some people prefer not to tell others of their problems and this choice must be respected. However, sometimes it can be reassuring and comforting to have someone to talk to, especially when your child has a long term chronic condition. Some families find comfort in religion and prayer.

It is important that you inform the brothers and sisters of your CF child

Try to explain to them the importance of the daily physiotherapy, enzymes and hospital visits. Always help them to understand the situation by answering their questions as best you can. Don't forget that they need your love and attention as well.

You may become very tired helping with treatment and running a home at the same time. Relatives, like grandparents, aunts and godparents can often help here. They may look after your affected child, or other children, when you are too tired or have other tasks to do. They may also be able to help with the physiotherapy.

It helps to confide in your nearest relatives and friends and tell them what CF is. You may even teach them to help you with the treatment. Without clearly understanding the disease, they may not understand the importance of physiotherapy or hospital visits. When your child looks healthy, they need to understand this is because of the correct treatment without which they would soon become ill.

If you feel that your relatives are not being helpful with their advice, or are trying to prevent you from carrying on with the treatment, it will help to persuade them to attend the CF clinic with you for a doctor to explain the situation to them.

You must make yourself strong to help your child to fight CF.

Treatment can be hard for the child and you may be tempted to give them a break from it. It is in your child's best interests that you *do not stop or decrease* the physiotherapy, enzymes, or hospital visits.

Apart from CF, your child is normal in every other way. A CF child will be expected to go through all the physical and mental experiences that a normal intelligent child goes through. They will get colds, sore throats, sports and games injuries like any other child irrespective of the fact they have CF.

Your child should be encouraged to be independent as it grows older and take on responsibility for its own treatment. This should happen gradually and the burden of the treatment should never rest entirely on your child's shoulders. Your child will not be able to do the treatment alone, especially when young. At 18 or 19 your child will be more independent but it is unrealistic to expect a child or adolescent to behave responsibly like an adult. The gradual transfer of responsibility for treatment will need to be carefully supervised. It is better for your child to be encouraged to accept responsibility rather than having it imposed. This way you will be able to work with your child to ensure all the treatments are carried out properly.

REMEMBER: CF is nobody's fault.

- Neither you, your child, or anybody in the family should feel any shame about it.
- If the child feels ashamed, they will try to hide it from their friends and from everybody else. They may stop taking the enzymes, and feel embarrased to cough in public.
- Your child should feel loved as much as any other child.

YOUR CHILD IS SPECIAL. You are not alone

For more information contact:

Austria: Austrian CF Association, Himmelreichweg 8, A-6112 Wattens
Belgium: Assoc. Belge de Lutte contre la Muscoviscidose,
Belgische vereniging voor strijd tegen Muscoviscidose, J. Borlélaan 12, 1160 Brussels, Belgium
Bulgaria: Cystic Fibrosis Assoc. of Bulgaria, Research Institute of Pediatrics,
Medical Academy, D. Nesterov str.II, 1606 Sofia, Bulgaria
Czechoslovakia: The Club of Parents and Friends of Children with CF,
Bitouska 1226/7, Praha 4 140 00 Czechoslovakia
Denmark: Danish Cystic Fibrosis Association, Hydrebakken 246, DK-8800 Viborg, Denmark
Estonia: Estonian Cystic Fibrosis Society (ECFC) 23 Riia St. Tartu, EE2400, Estonia
Finland: Association for the Pulmonary Disabled, Rehabilitation Secretary Seija Mauro,
PL 35-00620 Helsinki (Finland)
France: Association Française de Lutte contre la Mucoviscidose, 76, rue Bobillot, 75013 Paris, France, *et* SOS Mucoviscidose, ZAC de la Bonne Rencontre, 1 voie Gallo-Romaine,
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Greece: Hellenic Cystic Fibrosis Assoc., Parashou & Papathimiou Str. No.6, Athens 11475, Greece
Hungary: CF Foundation, H1124 Burok-u 15, Budapest, Hungary
Iceland: Cystic Fibrosis Assoc., of Iceland, Barnaspitali Hringsins, Landspitalinn v/Baronsstig,
101 Reyjavik, Iceland
Ireland: Cystic Fibrosis Assoc. of Ireland, CF House, 24 Lower Rathmines Road, Dublin 6, Ireland
Israel: Israel Cystic Fibrosis Assoc., 5 Shderot Hayeled, Ramat Gan, Israel 52444
Italy: Lega Italiana delle Associazioni per la lotta alla Fibrosi Cistica, presso Ospedale Civile Maggiore, Piazzali A. Stefani 1, 37126 Verona, Italy.
Luxembourg: Association Luxembourgoise de Lutte contre la Mucoviscidose asbl (ALLM),
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Switzerland: Schweizerische Gesellschaft für Cystische Fibrose (Mucoviscidose),
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