CYSTIC FIBROSIS MANAGEMENT AT PUBERTY AND THE PROBLEMS OF ADOLESCENT – ADULT TRANSITION PERIOD

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Abstract

Cystic fibrosis is the most frequent monogenic, authosomal recessive disease in Caucasian population. The complexity of this disease involves a special management fitting to every age and to clinical-evolutive moment. In the first part, the paper refers to specific aspects of cystic fibrosis management in teenagers about: the improvement of mucosal-secretion clearance, the control of infection, the maintaining of an optimal nutritional status, the prophylaxis and treatment of other diseases and complications of CF, the psychological support of the patient and his family. In the second part are presented the problems about the teenager-adult transition period, underlining psycho-affective aspects of the moment and care team role.

Keywords: cystic fibrosis, teenager, adolescent, care

Aim importance

Cystic fibrosis (FC) or mucoviscidosis is the most common autosomal recessive monogenic disease in the Caucasian population with chronic, progressive, potentially lethal evolution; a newborn from 2000-2500 is affected and one in 25 individuals is the carrier of the pathological gene [1,2]. In the absence of an early diagnosis, from the newborn period or at the earliest in the first years of life and from the right treatment applied, the chances of survival do not exceed the pre-school age. On the contrary, early diagnosis and correct management of the disease can assure an optimal quality of life for a long time [3].

At the moment, the life expectancy and quality of life of the patient with FC have improved significantly first of all due to acquisitions in disease awareness (after the discovery of the gene and defective protein) and, on the other hand, by developing a system complex treatment and care. FC is a complex and expensive disease, especially when we talk about an advanced development, and therefore any measure to prevent the progression of the disease must be applied without any reservation [3,4].

An important proof of the general interest in CF issues is the development of international programs, like the Cystic Fibrosis Worldwide Association (CFW), for the implementation and development of FC services in developing countries [4]

All these have led in the last decades to substantial changes in the mean survival of patients with FC [5,6]. If until 1970 the average of survival was 16 years, now has reached 32 years. In the same sense, it is estimated that for those born in the early 90's, the average age of survival will be over 40 years.

In this context, there are new problems of care, specific to older ages, also for the adult, many of the complications of the disease being related to the evolution of the disease in time (biliary cirrhosis, multifocal, diabetes, reproductive problems etc.)[3,4].

Cystic fibrosis management in puberty.

The development of services regarding FC management also in our country led to the improvement of the early diagnosis of the disease, even from the newborn period, todler and small baby. In the same time, the proper therapy, start at the installation of major complications, has determined, in time, an increasing rate of survival in patients with CF also in our country [4]. This makes the number of pubertal and adolescent patients currently increasing [7], which leads to new issues related to the management of the disease, including the transition to adulthood. In this context, home care therapy is the decisive component for ensuring a long-lasting quality of life for the patient with FC. Major goals in FC treatment are [3,8]:

1. Improvement of mucosal-secretion clearance
2. Control of infection
3. Maintaining of an optimal nutritional status
4. Prophylaxis and treatment of other diseases and complications of CF
5. Psychological support of the patient and his family

1.Improvement of mucosal-secretion clearance [9]

It is very important in the treatment of FC. The most sophisticated antibiotic treatment will be little or no effective in the absence of consistent and accurate physiotherapy. Physiotherapy has several components:
- airway clearance techniques;
- exercise;

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- mucolytic agents (aerosolotherapy): N acetyl cystine, Dornase alpha (breakes the leukocyte cytoplasmic DNA released from the local neutrophil - bacterial conflict, and which significantly accentuates the viscosity of the sputum);
- bronchodilators: since puberty, most CF patients develop bronchial hyperreactivity, bronchodilators (nebulised) become a standard component in the treatment of the disease.

2. Control of infection

When we talk about control of infection in FC is very important to know that there is so-called "point of no return" [10]. The infection once installed, especially with Pseudomonas aeruginosa, can no longer be eradicated, pulmonary lesions having a progressive evolution. Practically, infection control is achieved by antibiotherapy and anti-inflammatory therapy.

The principles of antibiotherapy are the following[11]:
- doses of drugs administered are higher than in other infections;
- the treatment is guided by the severity of the symptomatology and the causal germ (the germs involved in FC, especially Pseudomonas ae., do not respond to "usual" antibiotics, and it is recommended to use the antibiotics recommended by international working groups)
- the period of treatment will be prolonged, up to 3-4 weeks, including intravenous use.
- The administration of antibiotics nebulised will be done in prolonged months, even permanent [12].

Classical anti-inflammatory treatment refers to corticotherapy, reserved for advanced forms of disease, with high levels of circulating immune complexes. Administration is done in aerosols and / or oral, but with questionable benefits. Recent research has revealed a low level of essential fatty acids with an anti-inflammatory role despite good nutritional status, considering this aspect as specific for FC, probably related to the same defective gene [13]. Consequently, administration of essential fatty acids is regarded as an effective option in the anti-inflammatory treatment of FC.

3. Maintaining of an optimal nutritional status [14,15]

Maintaining an optimal nutritional status is aimed once for the compensating for pancreatic insufficiency by administering enzyme supplements exclusively in the form of minimal microspheres and on the other hand providing adequate nutrition according to certain standardized principles. Two aspects are to be taken into account in nutritional management:
- nutrition status monitoring through regular evaluations;
- nutritional intervention, generally reserved for two special situations: psychogenic context and / or exacerbation of pulmonary suffering: it can be achieved by: feeding supplements (Fresenius, Creacon) or by enteral nutrition (oro-nasogastric probe, gastrostomy, jejunostomy)

4. Prophylaxis and treatment of other diseases and complications of CF [16]

4.1. The equivalations of the meconial ileus (distal intestinal obstruction syndrome), a very common situation at older ages, is responsible for mechanical obstruction and emergency surgery. Supplements of pancreatic enzymes and gastrographic enema (under hospital conditions) are beneficial.

4.2. Chronic pancreatitis, for which the risk is very high in patients with a pancreatic phenotype. Monitoring of pancreatic amylases and lipases is mandatory after 10-12 years of age.

4.3. Gastroesophageal reflux, with no greater incidence in puberty and adolescence, should be considered, leading to major discomfort during physiotherapy.

4.4. Hepatobiliary suffering is clearly evident in this age group, translating into different clinical forms, from moderate hepatomegaly with fatty loading to millifocal biliary cirrhosis. Prophylactic treatment involves the administration of ursodeoxycholic acid in prolonged cures for years.

4.5. Decreased glucose tolerance and diabetes mellitus. The end of puberty and the beginning of adolescence represent, statistically, the periods of life when the glucose tolerance decreases and then the onset of diabetes. This evolutionary event has dramatic consequences, being responsible for the marked deterioration of pulmonary function and body mass index (17). Therefore, OGTT screening is mandatory after 10-12 years of age.

4.6. Associated osteopathy [3,21] . Recognized as a possible complication in the late `70s is defined by lowering bone density by more than 2 DS of the Z score or by the presence of one or more pathological fractures. Cross-sectional studies have revealed a high rate of fracture in MV patients compared to control groups, particularly in patients on waiting lists for transplantation. Pathogenesis is not fully elucidated. Morphohistometry studies in patients with stabilized disease have revealed a reduction in bone density, by reducing bone remodeling mechanisms and generally by altering the structure of connective tissue in the skeleton. Bone density assessment should be done by dual X-ray absorptiometry (DEXA). This investigation should be considered as part of the evaluation in all CF patients over the age of 6 years.

5 Psychological support of the patient and his family [18]

The complexity of the disease implies an extremely important role of psychological support in the evolution of the disease. This is addressed to the patient, his / her family and caregivers. The ways of psychological "intervention" adapt to the specifics of the disease, the psychology problems specific to the puberty period, adolescence, but also to the clinical-evolutionary moment. Generally, during the puberty period, the issues that occur in the evolution of the disease and which may influence the patient negatively are: Pseudomonas aeruginsa suprainfection, complications, increased number of hospitalizations, and decline in pulmonary function. Most frequently we see puberty in non-compliance reactions to treatment, especially physiotherapy [9].). It is time to apply the newer methods of physiotherapy (flute therapy, PEP mask), which shortens the time allocated

and creates some independence for the sick, may be beneficial.

Problems about the teenager-adult transition period, underlining psycho-affective aspects of the moment and care team role. [19,20]

Growing up, the teenager faces new medical and social problems. The transition from adolescence to adulthood is more difficult for these chronic patients because:
- must gain its affective and medical independence;
- to find a career path;
- to face the reality of the lack of radical treatment.

It's up to pediatricians and internists to discuss and consider all aspects of this transition. The care team must be complemented by pneumologist, gastroenterologist, surgeon, gynecologist, endocrinologist (diabetologist).

The transfer from the pediatric to the adult network around the age of 18 should be done in an optimal period (maturity, accept, except for periods of exacerbation of lung disease). The transition model depends on multiple factors:
- national and institutional health policy;
- financial possibilities;
- the proximity of pediatric and adult services;
- collaboration between pediatric and adult services.

In this context, the transition can be accomplished directly but with severe affective affects for the patient (not recommended) or progressively by overlapping for a period of time between the two pediatric and adult groups, the pediatrician being the coordinator of the two groups. The interdisciplinary care team must be a supportive professionalism because the quality of life is as important as its prolongation. The goals of adolescents / adults with FC exceed the goal of staying alive. They want independence, social and professional success and want to take responsibility for their lives.

Conclusions

The couple FC disease - is an unwanted marriage and without divorce, with implacable destiny, a life-long and dying battle. That is why, in order to not lose the fight from the start, the disease should not be considered an unbeatable enemy, but an adversary, which unfortunately still requires the rules. If the patient learns and obeys the rules, then his opponent (the disease) will also respect him. And sometimes, your opponent can become a partner, from whose evil one learns patience, dignity, tolerance and tenacity.

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