Journal of Case Reports: Clinical & Medical

Special Issue Article "Cystic Fibrosis"

Case Report

# Possible Consequences on the Patient because of the Psychological Impact and Parental Denial in the Diagnosis of Cystic Fibrosis: A Case Report

**Petrova G*, Perenovska P, Issaev V and Shopova S**

Pediatric Clinic, UMHAT "Alexandrovska", Medical University Sofia, Bulgaria

**ARTICLE INFO**

Received Date: November 22, 2018
Accepted Date: January 07, 2019
Published Date: January 08, 2019

**KEYWORDS**

Cystic fibrosis
Gastrointestinal tract
Psychological impact



**Citation for this article:** Petrova G, Perenovska P, Issaev V and Shopova S. Possible Consequences on the Patient because of the Psychological Impact and Parental Denial in the Diagnosis of Cystic Fibrosis: A Case Report. Journal of Case Reports: Clinical & Medical. 2019; 2(1):126

**Corresponding author:**
Petrova G,
Pediatric clinic, UMHAT "Alexandrovska", Medical University Sofia, Bulgaria,
Email: gal_ps@yahoo.co.uk

## ABSTRACT

Relationship between parents and children is unique for its endurance, responsibility, delicacy and vulnerability. Facing a chronic, incurable disease diagnosis in child sometimes could be unacceptable by parents.

We present a clinical case of a girl with diagnosed cystic fibrosis as a toddler, but failure in maternal coping with the diagnosis lead to significant progression of the disease and severe, irreversible changes in the lungs (multiple saccular bronchiectasis and lung function as low as 13% from predicted normal values). Besides lung damage, the patient also have marked failure to thrive, with a low body mass index and liver function also affected by the disease. The possible option for the patient could be lung transplantation, a procedure which hasn't been performed in Bulgaria and probably would not be possible in near future. Thus, the years, mother lost during her denial stage could be in fact fatal.

Stigmatizing a child with lethal disease could lead to strong denial of acceptance from the parents. In similar cases multidisciplinary approach to team of pulmonologist, gastroenterologist, dietician, geneticist, microbiologist and psychologist involved is crucial for a better outcome for both the patient and the family.

## INTRODUCTION

Cystic fibrosis (CF) is a complex, autosomal-recessive disorder, affecting the functions of respiratory system, gastrointestinal tract and all exocrine glands [1]. It's the most common fatal inherited disease in Caucasian affecting about one out of every 3,000 newborns in Europe [1] and 1 in 4000 in USA [2]. The chronic infections (mainly due to *Staphylococcus aureus, Pseudomonas aeruginosa and Burkholderiacepacia complex*) and inflammation, leading to bronchiectasis and respiratory failure are the leading cause of morbidity and mortality. In the last decades, survival of CF patients has been drastically improved due to newer therapeutic regimens, but still there is no a definite cure. Although with significantly improved survival and quality of life, compared to data 20 years ago, the cost for this achievement is substantial for both society and the patients [3]. Daily adequate treatment is very costly and time-consuming. It usually consists of more than 30 pills per day (including enzymes, vitamins, macro-and microelements, antibiotics, antifungal, liver protectors, anti-inflammatory and antacids), multiple nebulized sessions (with mucusthinningagents, antibiotics, bronchodilators and corticosteroids) and hours of respiratory cleaning techniques [4]. Frequent hospital visits also pay their negative role. Every chronic disease requires good compliance with treatment for the illness to be under

control. Non-compliance can lead to unmet treatment expectations and possibility for faster health decline and premature death [5]. Average rates of compliance in patients with chronic illnesses and particularly CF are from 50 up to 100% [6-10]. The more complicated the treatment is the lower compliance is [10].

Children with chronic illnesses are at increased risk (10-37%) of the development of psychological problems and coping of the child depends on parents' coping. Elisabeth Kubler- Ross describes emotional states experienced by patients after diagnosis of a chronic disease [11]. The five stages represent the normal range of feelings people experience when dealing with difficult changes in their lives. The first reaction is denial. In this stage individuals believe the diagnosis is somehow mistaken, they check the opinions of other doctors. Even if the test results are proven, patients tend to look for an error. When the individual recognizes that denial cannot continue they become frustrated – this is the anger stage. Psychological responses in this stage are directed towards looking for finding someone guilty. Patients at this stage often blame the doctor. The third stage involves the hope that the individual can avoid difficult situations. People can bargain or seek a compromise. During the fourth stage (depression), the individual is resigned, has no strength and lose hope. In this stage the patient may refuse meeting with people. Acceptance is the last stage when the patient accepts the situation and mobilizes all available resources for overcoming the disease. It is a time when the patient begins to cooperate in the treatment.

Relationship between parents and children is special. The parents play a substantial role in shaping children's emotional health, particularly in early childhood. Parents function as both a secure place for their child to discover their surrounding world and safe place in which they can return to the comfort and safety when they experience fear or discomfort. To properly fulfil this social role the parents have to overcome all different possible emotional and financial stress. The stress related to chronic, incurable disease in the child, sometimes could be unacceptable [12].

We present a clinical case of a girl born at term (birth weight 2800 g) from normal pregnancy with moderate asphyxiation, intrauterine infection and anaemia. Postpartum she required oxygen support, antibiotics and blood transfusion. In infancy the baby produced very smelly fatty stools – up to 10 daily, especially after greasy food and she presented with failure to thrive and bloated belly. At 2 years of age, she had prolapse of the rectum. In relation to these symptoms at 2 years and 10 months of age, she was diagnosed with cystic fibrosis by two positive sweat tests and genetic conformation (compound heterozygous for F508del, c.1521_1523delCTT, p.Phe508del and R1162X, c.3484C>T,p.Arg1162X). She was prescribed pancreatic enzyme replacement therapy (PERT), nutritional support, vitamins and ursodeoxycholic acid by the gastroenterologists. These two mutations from class I and class II are usually linked to a severe phenotype of the disease, which requires a multidisciplinary team approach and complex treatment [13]. Nevertheless the mother refused the specific therapy as she found every medication either 'not working' (PERT) or 'causing side effects' (i.e. purpura on the legs due to ursodeoxycholic acid). Since the girl had respiratory infections once yearly the mother entered into denial stage and she refused any therapy for CF claiming that the kid has lactose intolerance (due to some common symptoms in both diseases, but the child does not have confirmed lactose intolerance). A strict non-lactose dietetic regime with moderate effect (according the mother) was followed. The family was offered psychological help, which was refused by them at that time. Maybe due to the fear and incorrect beliefs in Bulgaria that the psychologist is equal to the psychiatrist, and only "crazy" people need psychological help. The father as the money maker for the family is hardly involved with the direct medical care of the patient. On the contrast of the mother, the father had accepted the diagnosis, but supported his wife's beliefs for maintaining a peaceful family atmosphere (like 'I agree with everything my wife says (even though I know she is wrong)').

Since the girl turned 4 years she started to cough regularly, and was treated by general practitioner for recurrent common cold. During this period, for unclear reason, no microbiology tests or chest X-ray was performed. First pulmonary exacerbation for which the family sought respiratory specialist occurred when the patient was 7 years and she was admitted in our clinic. At this first admission notable findings included: weight and height below 5th percentile (BMI of 14.3), decreased forced expiratory



Possible Consequences on the Patient because of the Psychological Impact and Parental Denial in the Diagnosis of Cystic Fibrosis: A Case Report. Journal of Case Reports: Clinical & Medical. 2019; 2(1):126.

volume for 1 second ($FEV_1$%) at 56.1% of predicted, sputum microbiology with *Staphylococcus aureus* and *Stenotrophomonasmaltophilia*, increased more than twice from the normal transaminases, slightly enlarged spleen, enlarged liver with homogenous hyperechogenic structure and cranio-caudal dimensions over 13 cm. The mother refused liver biopsy so liver cirrhosis was not histologically confirmed. The X-ray of the wrists showed 2 year delay in the bone age. The X-ray of the lung is shown in figure 1.

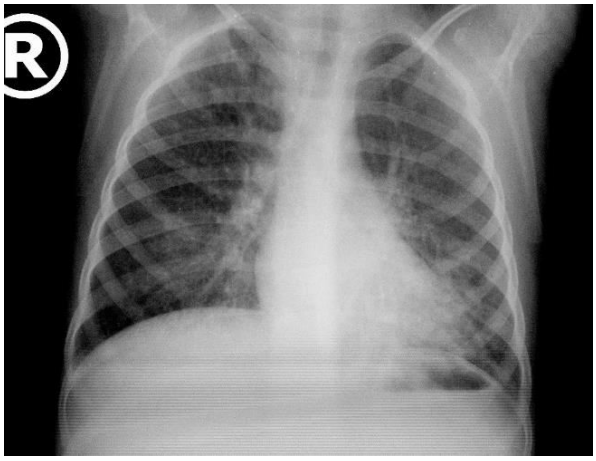

Figure 1: Initial X-ray of the patient (age 6 years and 11 months) – signs for bronchial obstruction in the right and multiple bilateral fibrosis changes.

FEV1% is a standard measure of pulmonary function used for assessing patient prognosis and several screening tools were developed combining FEV1% with other clinical factors [14]. Another risk factor related to poor prognosis besides low FEV1% is chronic infection with Staphylococcus aureus, Pseudomonas aeruginosa, Burkholderiacepacia complex or Mycobacterium abscessus [15].

The psychological evaluation showed normal neuro-psychological development of the patient, who was not informed about the disease. The child exhibited fear and anxiety (mostly hospital connected). The mother is overprotective and resistant towards the diagnosis and therapy. Despite the evident need for psychological help, the family refused to go to psychological family therapy. After discharge the mother refused any respiratory therapy and airway clearance techniques (ACT) because, the girl coughs too much from them ". The only medications she agreed to give were paroral antibiotics, according the sputum sample results (mainly for isolated *Staphilococcusaureus* and *Achrombacter xylosoxidans*). Almost 16 months later second pulmonary exacerbation in the clinic revealed also *Burkholdreiacepacia complex* infection. It was isolated for the first time in the patient. After 21 days of rigorous, combined intravenous antibiotic treatment, according to the antibiogram, the B.complex was not isolated in any subsequent sputum sample. The lung function was as bad as in the previous hospitalization period (X-ray of the lungs could be seen in figure 2). The family refused to see any psychologist at that time. The only thing they agreed afterwards was to do some ACTs (from time to time) and inhalation of dornase alfa 2-3 times a week. Bargaining with the idea that maybe her child has CF and if she was doing some ACTs they could avoid being in hospital. Although the mother seemingly agreed the presence of the disease, she still was in a denial stage since she was sure the child will 'outgrow' the disease.

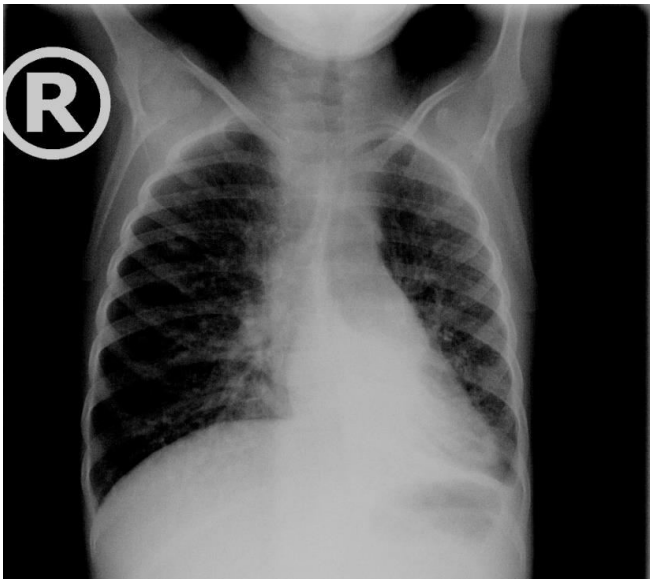

Figure 2: Second X-ray of the lung (age 7 years and 4 months) – more notable fibrosis changes bilaterally.

A year later the child was admitted with severely impaired general condition in orthopnoic position, TcSatO2 51%, p 150, grayish skin, cyanotic with severe mixed dyspnoea with total retractions and painful cough with thick green expectoration with FEV1 – 13%. The imaging studies (Figure 3 and 4) confirmed severe, irreversible changes in her left lung – signs of long-term non-compliance with the therapy. Mother was bargaining, offering even unimaginable things for her



Possible Consequences on the Patient because of the Psychological Impact and Parental Denial in the Diagnosis of Cystic Fibrosis: A Case Report. Journal of Case Reports: Clinical & Medical. 2019; 2(1):126.

child to survive. The next day when the child was a bit more stable, she was angry with the doctors blaming them for all health problems of the child including acquisition of 'strange' bacteria as Burkholderia or Stenotrophomonas. While we agree there is an increasing rate observed in many countries regarding the contagion of Burkholderiacepacia precisely in hospitals [16,17], this could not be valid for our patient. She was the first patient with isolated Burkholderia cepacia in our clinic for more than 6 years.

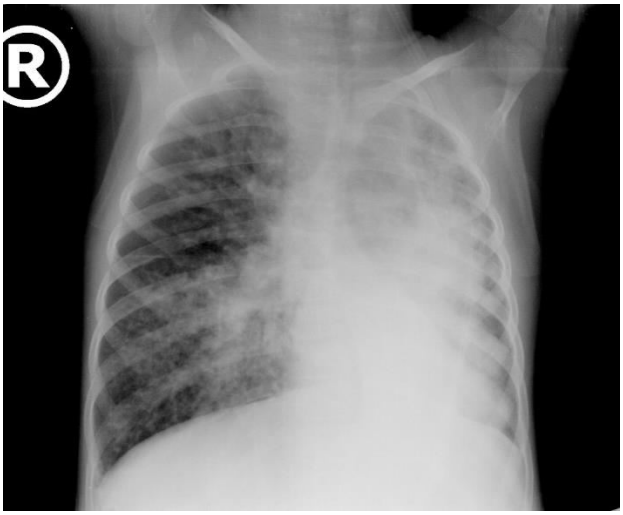


Figure 3: Third X-ray of the lung (age 8 years and 6 months) - The left lung field is almost homogeneously overshadowed. The right lung field has significantly increased transparency, right lung herniation in the left.

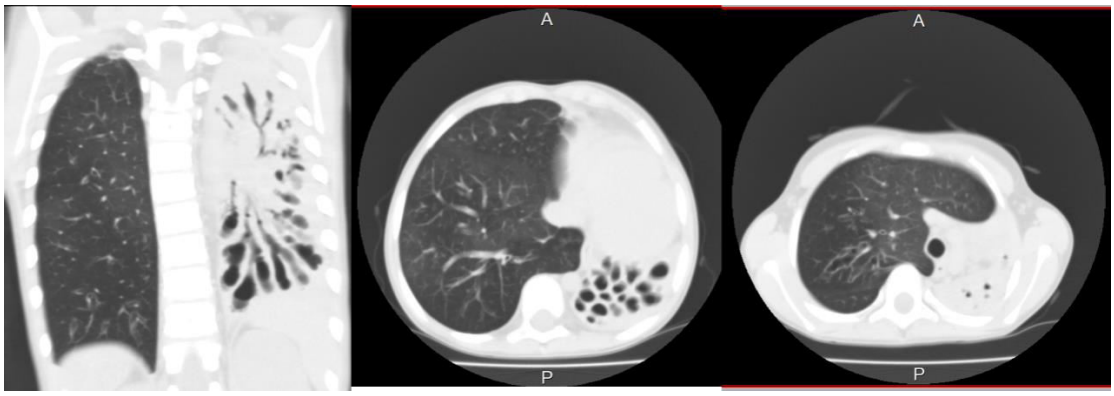


Figure 4: CT of the chest.

At least at that moment after the girl was successfully discharged at home the mother accepted the diagnosis of CF and agreed to follow the strict CF regimen.

Figure 4 CT of the chest (the same time as figure 3) – Not visible pulmonary parenchyma in the left, where highly over dilated segmental and subsegmental bronchi were found. The loss of volume in the left lung has led to the displacement and rotation of the entire mediastinal "shadow" so that the contour of the left of the heart is directly placed to the left thoracic wall distal to truncus pulmonalis which is expanded up to 22 mm. There is also visible emphysema of the entire right lung, the medial contour of which also reaches the left chest wall.

It took more than 7 years for the mother to start to accept the diagnosis, but these lost years of proper therapy had taken its



Possible Consequences on the Patient because of the Psychological Impact and Parental Denial in the Diagnosis of Cystic Fibrosis: A Case Report. Journal of Case Reports: Clinical & Medical. 2019; 2(1):126.

toll – since there are severe, irreversible changes in the lungs and the possible option for her could be lung transplantation. A procedure which hasn't been performed in Bulgaria and probably would not be possible in near future. This long period could be explained by the mother's own history of having had multiple pneumonias as child, very salty sweat and smelly stools at least 4 times daily, especially after greasy food. The mother denies the need for a sweat test or complete gene sequence in her, for she "doesn't want to feel guilty passing her child an incurable disease" if she turns out to have atypical CF herself. After the mother accepted the diagnosis, the patient's condition is relatively stable – not requiring any in hospital treatments or intravenous antibiotics for the last 3 months. The parents are fully motivated to seek also help in abroad in order the girl to be evaluated and if possible transplanted in the near future.

Noncompliance is a threat to the course of treatment, increases unnecessary diagnostic procedures, and confounds evaluation of effectiveness [11]. Factors related to compliance have been identified with regard to certain patient and disease characteristics, amount of support in the immediate environment, and the nature of the doctor-patient relationship. We have found that compliance is more difficult in families with low education or socioeconomic status, therefore our case was a real surprise for the family is well educated and has no financial burdens [18].

For the last 15 years, our team has consulted over 60 families with CF using anxiety scale tests, stress measurement when they receive the diagnosis for the child. We evaluated their coping strategies and their close and social relationships. We always prompted the families to seek a psychological consultation. Almost 80% of the families express increased anxiety when the diagnosis is confirmed. Only 1/3 (33% of all families) overcome the shock of diagnosis, accept and adapt to it through joint efforts and transparent relationships (mobilizing all coping resources). Almost 2/3 (60% of the families) have difficulties in adaptation to the diagnosis due to the low social competence, low integration and "masked conflict" in the family. About 7% of the families don't cope due to denying external and rapture of the internal communications, family connections crash (i.e. divorce, abdication of one of the parents, denying the diagnosis, searching for second opinion – for better diagnosis) [19]. The emotional trauma with coping with chronic illness [11,12,19] should not be labelled as a psychiatric disorder but as a transitory stage that requires psychological help. Despite the fatality of the disease, in good coping families the treatment burden is accepted and better clinical results are achieved [19].

In recent years, medicine has advanced significantly in terms of CF with a dramatic improvement in life expectancy. A child born today is expected to live more than 60 years, but this progress comes at the cost of severely intensified daily treatment that should not be interrupted [3]. The average patient with CF may need to take more than 30 tablets daily (antibiotics, vitamins, enzymes, liver protectors and nutritional supplements), repeated inhalations and physiotherapy morning and night just to keep their body in optimal condition [4]. This is associated with a loss of time and a lot of money, for in Bulgaria only PERT, dornase alfa and inhaled tobramycin/colomycin are free of charge and everything else is on patient/family expense. Everything is done to maximize preservation of organs and systems to the point where for each patient with CF will have adequate genetic treatment. As of 2018 there are three genetic modifiers on the market worldwide, however, none of them is available in Bulgaria, we can only hope in the near future our patients would benefit from such molecules. Children with CF are often at greater risk in understanding regimens because clinicians educate the parents and sometimes the difficulties in coping with the diagnosis prevent the option to pay a proper attention to this education, also sometimes the symptoms are misunderstood by both parent and general practitioner, and because of greater complexity of ACTs and a number of drugs and other aspects of treatment required. Stigmatizing a child with lethal disease could lead to strong denial of acceptance from the parents. In similar cases multidisciplinary approach with psychologist involved is crucial for a better outcome for both the patient and the family.

For the doctor has to „To cure sometimes, to relieve often and to comfort always"[20].

## REFERENCES

1. O'Sullivan BP, Freedman SD. (2009). Cystic fibrosis. Lancet. 373: 1891-1904.
2. Farrell PM, White TB, Howenstine MS, Munck A, Parad RB. (2017). Rosenfeld Metal. Diagnosis of Cystic Fibrosis in

Screened Populations The Journal of Pediatrics. 181: S33-S44.e2.

3. Simmonds NJ, Cullinan P, Hodson ME. (2009). Growing old with cystic fibrosis—the characteristics of long-term survivors of cystic fibrosis. Respir Med. 103: 629-635.
4. Smyth AR, Bell SC, Bojcin S, Bryon M, Duff A, et al. (2014). European Cystic Fibrosis Society Standards of Care Best Practice guidelines. J Cyst Fibros. 13: S23–S42.
5. Rafii F, Fatemi NS, Danielson E, Johansson CM, Modanloo M. (2014). Compliance to treatment in patients with chronic illness: A concept exploration. Iran J Nurs Midwifery Res. 19: 159-167.
6. Andal E. (2006). Compliance: Helping patients help themselves. Clin Rev. 116: 23-25.
7. Bissonnette J. (2008). Adherence: A concept analysis. J Adv Nurs. 63: 634-643.
8. Haynes RB, Yoa X, Degani A, Kripalani S, Garg A, et al. (2005). Interventions to enhance medication adherence (Review) Cochrane Database Syst Rev. 4: 1-77.
9. Conway SP, Pond MN, Hamnett T, Watson A. (1996). Compliance with treatment in adult patients with cystic fibrosis. Thorax. 51: 29-33.
10. Abbott J, Dodd M, Bilton D, Webb AK. (1994). Treatment compliance in adults with cystic fibrosis. Thorax. 49: 115-120.
11. Kubler- Ross E. (1969). On death and dying, Routledge. 10-26, 28,36
12. Petrova G, Shopova SI. (2004). Kalev Living with CF – Comparing the coping strategy in the families with CF-member and families with ALL-member J Cystic Fibrosis. 3: S102-S110.
13. CFTR2.
14. Bansal A, Mayer-Hamblett N, Goss CH, Heagerty PJ. (2017). A novel tool to evaluate the accuracy of predicting survival in cystic fibrosis. arXiv preprint arXiv. 1706.09602.
15. Lyczak JB, Cannon CL, Pier GB. (2002). Lung infections associated with cystic fibrosis. *ClinMicrobiolRev*. 15: 194-222.
16. Da Costa Capizzani CP, Caçador NC, Torres LAGMM, Tonani L, Vandamme P, et al. (2017). daCostaDarini AL Clinical and microbiological profile of chronic Burkholderia cepacia complex infections in a cystic fibrosis reference hospitalin Brazil. Eur J Clin Microbiol Infect Dis. 36: 2263-2271.
17. Mali S, Dash L, Gautam V, Shastri J, Kumar S. (2017). Anout break of Burkholderia cepacia complex in the paediatric unit of a tertiary care hospital, Indian J Med Microbiol. 35: 216-220.
18. Petrova G, Shopova A, Shopova S, Ouzunova I. (2005). Loneliness in the families of children with not so popular disease" J Develop Med & Ch Neuro. 4: S47.
19. Georgieva I, Shopova S, Hristozova I, Petrova G. (2006). "Coping Strategies of Families of Children with Life-Threatening Diseases (Malignant and Chronic Pulmonary). Comparative Analysis", *Pediatrics* magazine. 46: 23-26.
20. Kumar A, Allaudeen N. (2016). To Cure Sometimes, to Relieve Often, to Comfort Always. JAMA Intern Med. 176: 731-732.