Assessment of the biological condition and nutritional status of adult patients with cystic fibrosis

Magdalena Kosińska, Anita Szwed, Joachim Cieślik, Joanna Goździk, Karol Karbowy

1 Institute of Anthropology, Adam Mickiewicz University, Umultowska 89, 61-614 Poznań, Poland, E-mail: gdusia@amu.edu.pl
2 Chair and Department of Phtysiopneumonology, Academy of Medical Sciences, Szamarzewskiego 89, Poznań, Poland

ABSTRACT The aim of the study was to assess the biological condition and nutritional status of adult cystic fibrosis patients. The study included 55 individuals aged from 18 to 31 years. Their biological condition and nutritional status were determined by somatometric parameters and their pulmonary function by spirometry and a microbiological test. The results show considerable physical retardation and poor nutritional status of the studied adult cystic fibrosis patients. It was also shown that the type of mutation affects body mass and height, as well as thigh and calf circumference values, while the severity of the respiratory system disorder modifies only the body mass value (Kruskal-Wallis test: $H (3,55)= 8.505, p < 0.04$). The authors have demonstrated a statistically significant relationship between the type of mutation and severity of clinical respiratory symptoms, and nutritional status (respectively: $H (2,55) = 9.589, p < 0.01; H (3,55) = 13.320, p < 0.005$).

KEY WORDS Cole index, somatometric traits, cystic fibrosis mutation, pulmonary function, developmental retardation, malnutrition

Introduction Cystic fibrosis is the most frequent autosomal recessive monogenic disease. The frequency of this pathology in Poland is estimated at approximately 1:2500 births [BOŻKOWA et al. 1971]. Cystic fibrosis is incurable but owing to more effective treatment of respiratory system infections and an improvement in nutritional behavior over the last 20 years the clinical condition of the patients has improved considerably and mean life expectancy has increased considerably (in 1940 it was one year, in 1969 – 14 years and in 1995 it was estimated as 30 years) [YANKASKAS et al. 2004].
The cystic fibrosis gene (CFTR – Cystic Fibrosis Transmembrane Conductance Regulator) was located in the mid 1980s on the long arm of chromosome 7 [KEREM et al. 1990; WITT 1994]. Over 1000 mutations of this gene are currently known [Cystic Fibrosis Genetic Analysis Consortium]. The CFTR gene mutations, among which the ΔF508 mutation (a deletion of three nucleotides (CTT) resulting in the loss of phenylalanine residue at position 508 on the peptide chain) is the most frequent, cause CFTR protein dysfunction, and consequent impaired secretion of chloride ions by glandular epithelium cells [STRANDVIK 1991]. Exocrine glands produce viscous and thick mucus, with clinical sequelae in the form of recurrent pulmonary inflammations and sinusitis, exocrine pancreatic insufficiency, liver damage, as well as energy balance disorders. Another commonly observed disorder is retarded biological development of the patients manifesting as negative deviations of somatometric traits compared to healthy peer group values [e.g., BYARD 1990; LANDON and ROSENFELD 1984; MÜLLER et al. 1999]. The genesis of this growth retardation has not been unequivocally explained so far. Initially, the diversity of clinical expression and the degree of developmental deviations from the normal biological condition were explained by the fact of occurrence of many mutations of the CFTR gene. However, assuming that in the course of physical development an interaction occurs between the genetic determination and external environmental factors, one should expect the occurrence of developmental differences resulting from genotypic variation and from the modifying effect of the environment.

One of the fundamental, though still underestimated, problems in cystic fibrosis treatment is proper nutrition. In the past malnutrition and physical retardation were considered to be an inherent element of the disease process. However, as demonstrated by COREY et al. [1988] and LAI et al. [1999], a properly composed diet has a significant effect on the course of the disease and the patient's physical development. The nutritional status of cystic fibrosis patients is a crucial prognostic of the course of the disease, including also maintenance of respiratory system efficiency [REOSENFELD et al. 1997; HAYLLAR et al. 1997; COREY and FAREWELL 1995; GRASEMANN et al. 1995].

The goal of the study was to assess the biological condition and the nutritional status of adult patients with cystic fibrosis.

Materials and methods

The studied group consisted of 55 adult cystic fibrosis patients (24 males and 31 females), aged from 18 to 31 years (respectively: \( \bar{x} = 22 \pm 3.64; \bar{x} = 22 \pm 2.10 \)). The diagnosis of CF was established in accordance with the criteria proposed by the Polish Cystic Fibrosis Working Group [Zasady rozpoznawania cystycznego ... 2000] (one or more clinical symptoms of the disease present, family history of cystic fibrosis, positive result of the CF newborn screening test). Confirmation was obtained based on the perspiration test, measurement of transepithelial potential difference and molecular tests of the CFTR gene performed in the Human Genetics Institute of the Polish Academy of Sciences in Poznań.
The patients were divided into three groups of subjects according to mutation type. The first group (18%) were people with the ΔF508/ΔF508 genotype, classified as a severe mutation. The second group (67%) were people having a mutation described as mild, i.e., individuals with the ΔF508/M genotype, where M is the other “unspecified” mutation, causing a more benign form of the disease. The third group (15%) consisted of individuals with the M/M genotype. No statistically significant differences in the frequency of the specified mutation types were found between sexes ($\chi^2_{df=2} = 0.256; p > 0.05$).

The assessment of biological condition and nutritional status was performed on the basis of somatometric traits. Measurements were taken according to the techniques introduced by Martin and Saller in 1957 [MALINOWSKI and BOŻIŁOW 1997]. The measurements were performed using Swiss GPM anthropometric instruments and electronic scales. Circumferences were measured using anthropometric tape with accuracy up to 1 cm. Thickness of skinfolds was measured with a skinfold meter with standard pressure of 10 g/sq. mm and accuracy up to 1 mm.

The degree of severity of the bronchopulmonary disease was assessed by spirometry and by the standard microbiological sputum test for the presence of Pseudomonas aeruginosa pathogen.

The collected data were analyzed statistically by both parametric and non-parametric methods used in compliance with their assumptions. Deviations of the subjects' physical development status from the healthy population were assessed on the basis of the single mean t-test. The assessment of the effect of mutation type and the degree of severity of clinical respiratory symptoms on the values of somatometric traits and on nutritional status was based on the ANOVA and Kruskal-Wallis ranks test. In the course of further analyses, multiple comparisons of rank means were performed in order to estimate differences between individual groups.

All values of the studied somatometric traits were standardized. Values of the traits in healthy male 18-year olds and female 18-year olds given in “Dziecko Poznańskie” [CIEŚLIK et al. 1994] and “Dziecko Konińskie” [1998] were used as reference. Measurements of skinfolds' thickness were presented in logarithmic form, using logarithmic transformation tables constructed by Edwards et al. [Dziecko Konińskie 1998]:

Transformation = 100 [log (skinfold in mm – 1.8) +1]

The values of the Cole index expressed in percent enable the following classification of nutritional status:

- $> 110$ — overnutrition
- $90 – 110$ — normal nutrition
- $85 – 90$ — slight malnutrition
- $75 – 85$ — malnutrition
- $75$ — emaciation

The statistical analysis of the material was performed using software package STATISTICA 7.0 (StatSoft. Inc. 2005).
Results

The physical development status of the subjects was assessed on the basis of somatometric traits. The results of the single mean \( t \)-test demonstrate that the physical development in individuals suffering from cystic fibrosis is considerably retarded compared to healthy individuals. The majority of the mean values of standardized somatometric variables exhibit significant statistical deviations in the negative direction compared to the healthy population (Table 1). However, one should note the high variability of the studied traits, for instance for body mass and height the minimum value exceeds minus 4 standard deviations, and the maximum value indicates that among the subjects there are also individuals whose body height and mass do not deviate from the values obtained for the healthy population (Fig. 1). It was also observed that in the studied group there was a higher percentage of individuals with body mass deficiency exceeding minus 2 SD (42%) compared to individuals with body height deficiency exceeding minus 2 SD (29%). However, the difference between the frequencies is not statistically significant (\( \chi^2 = 1.95; p > 0.05 \)). At the same time, it should be noticed that over 70% of the subjects have an extremely low (< -2SD) thigh circumference (Fig. 2).

A further analysis demonstrated a significant effect of mutation type on body mass and height values and thigh and calf circumferences (Table 2). In turn, the severity of the respiratory symptoms modifies only body mass values of the subjects (Kruskal-Wallis test: \( H (3,55) = 8.505, p < 0.04 \)).

A crucial aspect of the study was the estimation of nutritional status of individuals with cystic fibrosis. It was observed that almost 60% of the patients showed malnutrition symptoms, ranging from slight malnutrition to symptoms indicating emaciation. However, it should be noted that 4% of the subjects were classified into the excessive nutritional status category (Table 3). Nutritional status may be conditioned by many

<table>
<thead>
<tr>
<th>Somatometric trait (N=55)</th>
<th>( \bar{x} )</th>
<th>SD</th>
<th>( p )</th>
</tr>
</thead>
<tbody>
<tr>
<td>Birth body weight</td>
<td>-0.20</td>
<td>0.79</td>
<td>&gt; 0.05</td>
</tr>
<tr>
<td>Body mass</td>
<td>-1.58</td>
<td>1.21</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>Body height</td>
<td>-1.19</td>
<td>1.24</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>Triceps skinfold</td>
<td>-0.08</td>
<td>1.12</td>
<td>&gt; 0.05</td>
</tr>
<tr>
<td>Subscapular skinfold</td>
<td>-0.61</td>
<td>1.04</td>
<td>&lt; 0.01</td>
</tr>
<tr>
<td>Abdomen skinfold</td>
<td>-0.54</td>
<td>0.84</td>
<td>&lt; 0.01</td>
</tr>
<tr>
<td>Upperarm circumference</td>
<td>-1.70</td>
<td>1.15</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>Forearm circumference</td>
<td>-1.33</td>
<td>1.33</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>Thigh circumference</td>
<td>-2.84</td>
<td>1.13</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>Calf circumference</td>
<td>-1.74</td>
<td>1.27</td>
<td>&lt; 0.001</td>
</tr>
<tr>
<td>Chest circumference</td>
<td>-0.51</td>
<td>1.12</td>
<td>&lt; 0.03</td>
</tr>
<tr>
<td>Transverse chest breadth</td>
<td>-0.42</td>
<td>0.83</td>
<td>&lt; 0.02</td>
</tr>
<tr>
<td>Sagittal chest depth</td>
<td>0.42</td>
<td>1.10</td>
<td>&gt; 0.05</td>
</tr>
</tbody>
</table>
Fig. 1. Physical development of studied persons – normogram of the variability of somatometric traits.

Fig. 2. Standardized deviations of somatometric traits ($p < 0.05$) – frequency of occurrence.
The performed analyses demonstrated a statistically significant relationship between the mutation category and severity of the clinical respiratory symptoms, and nutritional status (respectively: $H (2,55) = 9.589, p < 0.01$; $H (3,55) = 13.320, p < 0.005$). In the course of further analyses, multiple comparisons indicated that individuals with the severe mutation and individuals with severe respiratory symptoms tended to have more clearly marked physical retardation and worse nutritional status ($p < 0.05$).

### Table 2. Effect of mutation type on somatometric traits

<table>
<thead>
<tr>
<th>Somatometric trait (N=55)</th>
<th>$H (2,55)$</th>
<th>$p$</th>
</tr>
</thead>
<tbody>
<tr>
<td>Birth body weight</td>
<td>1.110</td>
<td>&gt; 0.05</td>
</tr>
<tr>
<td>Body mass</td>
<td>12.535</td>
<td>&lt; 0.002</td>
</tr>
<tr>
<td>Body height</td>
<td>8.682</td>
<td>&lt; 0.02</td>
</tr>
<tr>
<td>Triceps skinfold</td>
<td>5.025</td>
<td>&gt; 0.05</td>
</tr>
<tr>
<td>Subscapular skinfold</td>
<td>2.454</td>
<td>&gt; 0.05</td>
</tr>
<tr>
<td>Abdomen skinfold</td>
<td>3.686</td>
<td>&gt; 0.05</td>
</tr>
<tr>
<td>Upperarm circumference</td>
<td>2.245</td>
<td>&gt; 0.05</td>
</tr>
<tr>
<td>Forearm circumference</td>
<td>5.715</td>
<td>&lt; 0.05</td>
</tr>
<tr>
<td>Thigh circumference</td>
<td>6.014</td>
<td>&lt; 0.05</td>
</tr>
<tr>
<td>Calf circumference</td>
<td>7.745</td>
<td>&lt; 0.03</td>
</tr>
<tr>
<td>Chest circumference</td>
<td>1.669</td>
<td>&gt; 0.05</td>
</tr>
<tr>
<td>Transverse chest breadth</td>
<td>2.845</td>
<td>&gt; 0.05</td>
</tr>
<tr>
<td>Sagittal chest depth</td>
<td>2.516</td>
<td>&gt; 0.05</td>
</tr>
</tbody>
</table>

### Table 3. Estimation of nutritional status of studied persons

<table>
<thead>
<tr>
<th>Nutritional status</th>
<th>N</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Overnutrition</td>
<td>2</td>
<td>3.64</td>
</tr>
<tr>
<td>Normal nutrition</td>
<td>21</td>
<td>38.18</td>
</tr>
<tr>
<td>Slight malnutrition</td>
<td>7</td>
<td>12.73</td>
</tr>
<tr>
<td>Malnutrition</td>
<td>15</td>
<td>27.27</td>
</tr>
<tr>
<td>Emaciation</td>
<td>10</td>
<td>18.18</td>
</tr>
</tbody>
</table>

### Discussion

In starting to discuss the obtained results one should point to the small size of the sample, which is typical of clinical studies. This fact results in certain interpretational constraints.

The physical development status and nutritional status of cystic fibrosis patients are affected by many factors. The causes of physical development and nutritional status disorders are explained in two ways. The first explanation assumes that deviations in this respect are a secondary consequence of gastrointestinal or respiratory disorders, conditioning negative energy balance and malnutrition. This results in the body's decreased energy output for growth and tissue regeneration at the expense of support of the vital organs' functions, and in a delayed physical development. In cases involving such changes regular therapy and a properly composed diet effectively improve the patients developmental status [GRABOWSKA and ŁUCZAK 1996]. According to the second explanation, the scale of developmental retardation coexistent with the disease is in fact independent of the degree of severity of disease and should not be treated as a secondary consequence of the pathological process’ complications. In light of this interpretation, developmental retardations are ones of the effects of pleiotrophy at the cystic fibrosis locus which may have an effect on the change of the original expression of the gene defect [SING et al. 1982; MAHANEY, MCCOY 1986].

The results obtained in this study, demonstrating a considerable physical retardation of cystic fibrosis patients
compared to the healthy population, are consistent with the results obtained by other authors [SHEPHERD et al. 1980; MEARN 1983; GRABOWSKA and ŁUCZAK 1996; SHEPHERD et al. 1986]. A significant shift of the mean values of traits in the negative direction was observed. However, it is worth noticing that within the studied sample includes both individuals with extremely low phenotypic values and individuals with values similar to the healthy population. The biggest deviations were recorded in body mass and height and thigh and calf circumferences. However, suggested by many authors [e.g., COREY et al. 1988; BARKHOUSE et al. 1989; LAI et al. 1999] predominance of body mass deficiency over body height deficiency was not observed. In fact, higher values of body mass deviations in comparison with height deviations are not a steadfast rule, since studies by BORBEL and DOCTTER [1997] and WALKOWIAK [1998] failed to demonstrate a predominance of body mass deficiency. The present results show a higher percentage of individuals with body mass deficiency exceeding minus 2 SD (42%) compared to individuals with body height deficiency exceeding minus 2 SD (29%). Nevertheless, the difference between the observed values is not statistically significant ($\chi^2 = 1.95; p > 0.05$). As demonstrated by LAI et al. [1999], similar low values have been observed in recent years only in 10-15% of cystic fibrosis patients. The results obtained in this study approximate those observed in highly developed countries in past decades. This is probably due to the fact that intensive nutritional management in Poland started to be implemented only a few years ago [NOWAKOWSKA et al. 1995].

The presented results indicate a significant shift of the means of standardized values of the studied circumferences in the negative direction, with the exception of the chest circumference. The observed deviations in relation to the healthy population agree with the literature data pertaining to children [GRABOWSKA and ŁUCZAK 1996]. The chest circumference being situated within the normal range for the population results from its barrel-like shape (increased sagittal depth with accompanying decreased transverse breadth) typical of this type of pathology.

Mean standardized values of a skin-fold thickness in the studied group do not deviate from accepted normal values for the healthy group of individuals, which is in agreement with the result obtained by BENTUR et al. [1996]. On the other hand, studies conducted by RICHARDSON et al. [2000] demonstrated a decreased thickness of skin folds in cystic fibrosis patients.

In their studies, LAI et al. [2000] and MEARN [1983] described birth weight deficiencies in newborns with cystic fibrosis, even after the exclusion of newborns of low gestational age and those with meconium ileus. In turn, BRONSTEIN et al. [1992] demonstrated significant deficiencies in body weight at birth only in girls. The authors suggest that deficient growth may start as early as the gestation period. According to the study by MÜLLER et al. [1999], considerable lowering of the birth body weight in newborns with cystic fibrosis (both girls and boys) may result from intrauterine malnutrition caused by pancreatic insufficiency in the fetus. However, the results obtained in this study demonstrate no statistically significantly low-
erred values of body weight at birth in individuals with cystic fibrosis as compared to values obtained for healthy individuals. Similar results were obtained by Greer et al. [1991] who found that birth body weight values in cystic fibrosis patients were within normal range.

As expected, the authors observed a significant effect of mutation type on body mass and height values and on thigh and calf circumferences. In patients with a severe mutation, physical retardation is more clearly marked. This mutation causes exacerbation of disease symptoms and, as a result, affects the overall biological condition of the patient. It is worth noticing, that this type of mutation has no differentiating effect on the dimensions of the chest. It transpires from the literature that despite the fact that changes in the chest, most likely caused by functional disorders of the respiratory system, are registered in as many as 95% of cystic fibrosis cases, they are dependent on the type of mutation [Kostuch and Wojcierowski 1995]. The presented analyses also failed to demonstrate a relationship between the formation of skinfolds and the type of mutation. This may mean that these traits are highly ecosensitive. A similar situation is observed with regard to the mean body weight at birth. This would corroborate the thesis that the reasons for the demonstrated differentiating effect of the type of mutation on certain traits (body mass and height, thigh and calf circumferences) should be looked for in a secondary effect of functional disorders of individual genotypically conditioned systems rather than among the direct effects of the genetic determination.

From abundant literature on the subject of cystic fibrosis it is known that the biological condition of cystic fibrosis patients is affected by clinical respiratory symptoms. However, the results obtained by the authors demonstrate a relationship only between respiratory manifestations and body mass. Similar results were obtained by other authors, e.g., Byard [1989] and Collins et al. [1999]. According to these researchers, lowered body mass is prognostic of pulmonary disease, and anthropometric parameters correlate more strongly with pulmonary function than with, for instance, parameters indicating pancreatic insufficiency. Research by Sproul and Huang [1964] also indicate significant relationships between exacerbation of respiratory symptoms and a lowered body mass.

The nutritional status is defined as a complex of somatic and biochemical body traits, being a manifestation of the supply of nutrients to the body, their digestion, absorption and metabolism. Resulting from their assessment of the nutritional status of the subjects, the authors were able to corroborate the state of malnutrition in cystic fibrosis patients as suggested earlier by many authors [Kerem et al. 1990; Ryzko 1997; Hayllar et al. 1997]. In the studied group, this ranged from slight to the level indicating emaciation. The obtained results show unequivocally that despite considerable advancement in the medical sciences and a better than before degree of health consciousness, patients with cystic fibrosis still demonstrate poor nutritional status. However, the existence of considerable differences among subjects in this respect, meaning
the presence of adequately nourished, and even overnourished individuals in the studied group, highlights the importance of more appropriate nutritional therapy and more efficacious medical care in the treatment of cystic fibrosis.

One cannot, however, neglect the importance of the effect of the type of mutation and severity of the respiratory disease symptoms on the nutritional status. Patients with a severe mutation type and those with an obvious pulmonary dysfunction were observed to have inferior nutritional status. No overnourished individuals were found in either the first or the second group. The correctness of the obtained results is confirmed by the fact that other pulmonary diseases (e.g., chronic obstructive lung disease) also result in malnutrition [SAHEBJAMI 1989]. It needs to be emphasized that there exists a strong relationship between the type of mutation and respiratory symptoms, and the nutritional status of cystic fibrosis patients. The more severe the mutation and respiratory symptoms, the worse is the nutritional status of patients with cystic fibrosis. The observed deviations of the anthropometric traits, in locating the subjects below the norm adopted for the healthy population, may result from both inhibited (delayed) growth and malnutrition. It is difficult to state unequivocally whether retarded development is a result of malnutrition or whether malnutrition is a result of inhibited development. Cystic fibrosis is a disease "with many faces" and it is thus difficult to ascertain cause and effect in the observed disorders. On the basis of various results presented in the literature, one can state that correlations of respiratory system functions with specific CFTR gene mutations are not unequivocal. Patients homozygotic for ΔF508 are more prone to chronic bacterial infections, being mostly Pseudomonas aeruginosa colonization [WITT 1994]. Only patients homozygotic for this allele are diagnosed with meconium ileus of the alimentary system as being a cause of malnutrition. It was also demonstrated that nonsense mutations had more severe negative effects on the gastric system than on the respiratory system, though causes of this phenomenon are still unknown. Further, patients with the same genotype were found to exhibit considerable differences in pulmonary symptoms [KEREM et al. 1990], which may be an argument in favor of the impact of other genetic factors or environmental modification in the expression of the disease.

Conclusions

The phenomenon of retarded biological development which is reflected in the developmental status of adult individuals is an inseparable element of many pathological processes. On the basis of the results obtained in this study, the authors ascertained that adult cystic fibrosis patients demonstrated considerable retardation in physical development and a poor nutritional status. The type of mutation and severity of respiratory symptoms significantly affect the physical development and nutritional status of cystic fibrosis patients.

Acknowledgements

We are most grateful to Prof. Michał Witt from the Human Genetics Institute of the Polish Academy of Sciences in
Poznań for molecular tests of the CFTR gene. We also thank the referees for their comments on the original manuscript of this paper. This study was supported by interdisciplinary grant founded by Adam Mickiewicz University and Academy of Medical Sciences in Poznań (No. PU-II/10).

References


Cystic Fibrosis Genetic Analysis Consortium, http://www.genet.sickkids.on.ca/cftr-cgi-bin/ Mutation


GRABOWSKA J., B. ŁUCZAK, 1996, Rozwój biologiczny dzieci i młodzieży chorych na mukowiscydozę, [in:] Antropologia a medycyna i promocja zdrowia, t.1, A. Malinowski (ed.), Łódź, pp. 142-154

GRASEMANN H., H.G. WIESEMANN, F. RATJEN, 1995, The importance of lung functions as a predictor of two year mortality in mucoviscidosis, Pneumonologie, 49, 466-469


Streszczenie

Mukowiscydoza jest najczęściej występującą autosomalną recesywną chorobą monogenową. Do najczęstszych objańców klinicznych mukowiscydozy zalicza się zapalenia płuc i żołądka, niewydolność zewnętrznych chłonników, cholestaz oraz podwyższenie poziomu elektrolitów w pocie. Powszechnie obserwowane jest również opóźnienie rozwoju biologicznego, a w przypadku osób dorosłych – niedobory masy i wysokości ciała.

Głównym celem badań była ocena stanu biologicznego oraz stopnia odżywienia osób dorosłych chorych na mukowiscydozę. Badaniami objęto 55 osób chorych (24 mężczyzn oraz 31 kobiet), w wieku od 18–31 lat. Rozpoznanie postawiono zgodnie z kryteriami zaproponowanymi przez Polską Grupę Roboczą Mukowiscydozy [Zasady rozpoznawania ... 2000]. Potwierdzenie uzyskano w oparciu o test potowy, test wysokich wartości przeznaczeniowej różnicy potencjałów oraz badania molekularne genu CFTR. Na podstawie rodzaju mutacji

Richardson I., I. Nyulasi, K. Cameron, M. Ball, J. Wilson, 2000, Nutritional status of an adult cystic fibrosis population, Nutrition, 16(4), 255-259
Walkowiak J., 1998, Stan odżywienia i rozwój fizyczny dzieci chorych na mukowiscydozę w świetle podstawowych wskaźników wagowych i wzrostowych, Przegląd Pediatriczny, 28(3), 208-212
Zasady rozpoznawania i leczenia mukowiscydozy, 2000, Stanowisko Polskiej Grupy Roboczej Mukowiscydozy przy Zarządzie Głównym Polskiego Towarzystwa Pediatricznego, Medycyna Praktyczna – Pediatria, 3, 64-74
wyróżniono trzy grupy badanych: 1) osoby o genotypie ∆F508/∆F508 uznawanym za mutację „ciężką” (N = 10; 18%); 2) osoby o genotypie ∆F508/M, gdzie M to inna mutacja – tzw. nieokreślona, powodująca łagodniejszy przebieg choroby (N = 37; 67%); 3) osoby o genotypie M/M (N = 8; 15%). Nie stwierdzono różnic istotnych statystycznie w częstości występowania wyróżnionych kategorii mutacji pomiędzy płciami ($\chi^2_{df=2} = 0,256; p > 0,05$). Stan biologiczny oraz stopień odżywienia określono przy pomocy parametrów antropometrycznych, a funkcję płuc – w spirometrri oraz w badaniu mikrobiologicznym płwociny na obecność Pseudomonas aeruginosa. Zebrane dane poddano analizie statystycznej obejmującej metody parametryczne i nieparametryczne, zastosowane zgodnie z ich założeniami. Wszystkie wartości badanych cech antropometrycznych zstandaryzowano. Pomiary grubości falców skóro-tłuszczowych przedstawiono w postaci logarytmicznej, wykorzystując tablice przekształceń logarytmicznych dla tych pomiarów, skonstruowanych przez Edwardsa i wsp. [Dziecko Konińskie 1998].

Uzyskane wyniki wskazują, że rozwój fizyczny osób chorych na mukowiscydozę jest znacznie opóźniony w stosunku do osób zdrowych. Średnie wartości standaryzowanych zmienności antropometrycznych wykazują istotne statystycznie odchylenia w kierunku ujemnym w stosunku do populacji zdrowej (tabl. 1, fig. 1). Typ mutacji wpływa na wartości masy i wysokości ciała oraz obwodów uda i podudzia (tabl. 2). Nasilenie dolegliwości ze strony układu oddechowego modyfikuje jedynie wartość masy ciała (test Kruskala-Wallisa: $H (3,55) = 8,505; p < 0,04$). Osoby dorosłe chore na mukowiscydozę wykazują znaczne opóźnienie rozwoju fizycznego i zły stan odżywienia. Wykazano istotną statystycznie zależność pomiędzy kategorią mutacji i nasileniem objawów klinicznych ze strony układu oddechowego a stanem odżywienia (odpowiednio: $H (2,55) = 9,589; p < 0,01; H (3,55) = 13,320; p < 0,005$).