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Frequency Distribution and Association of some Morpho- and Physiological Traits in Patients with Lung Diseases in Kosova

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ABSTRACT

The aim of this study was to investigate the distribution of specific phenotypes in patients with lung diseases as well as their eventual association with the risk of developing lung diseases. For this purpose 2777 patients with lung diseases and 2778 healthy individuals from all over Kosova were examined for the appearance of the following selected phenotypes: ear lobe free (ELF)/ ear lobe attached, normal chin (NC)/cleft chin, tongue roller (TR)/non roller, hand clasping right thumb over (HC)/ hand clasping left thumb over, righthanded (RH)/lefthanded. In addition, the blood group from ABO system and the presence or absence of the Rhesus factor as phenotypical markers were observed. The results obtained show significant differences between control and lung disease patients for NC ($p \leq 0.05$) and TR ($p \leq 0.005$) as well as for blood groups AB ($p \leq 0.05$) and O ($p \leq 0.005$). These results point to eventually increased levels of genetic load as a result of the increased homozygosity in some gene loci causing an increased frequency of some recessive phenotypes in patients with lung diseases. Together with the specific associations observed, these preliminary findings could serve as a basis for further in depth investigations with respect to the types of lung diseases, occupational exposure and dietary habits, and thus is expected to contribute to an understanding of predispositions and susceptibility to lung diseases.

Key words: *lung diseases, phenotype, patients, susceptibility, Kosova*

Introduction

The investigation of the mechanisms that generate and maintain genetic variation in populations is one of the main topics in population genetics. In these investigations, different mathematical models are used to describe the distribution/frequencies of certain features as an early prediction providing solid support to carry out further studies on genetic epidemiological associations[1-4].

Analysis of variability of qualitative traits is still one the most suitable approaches to verify the differences and similarities between populations of organisms. About 3000 recessive autosomal features are listed in »Online Mendelian in Man« database[5] representing the second largest group after those inherited in an autosomal dominant way. Besides the »benign« recessive features which for example determine the inability of tongue rolling, attached ear lobes etc., genetic conditions responsible for cystic fibrosis, spinal muscular atrophy, phenylcetonuria and Tay-Sachs disease are also of autosomal recessive nature[6]. Since the 30s and 40s of the last century it was argued that natural populations of different types carry higher proportions of recessive micromutations called »genetic load«[7] and stated that recessive homozygotes are in general more prone for diseases (especially for rare ones, e.g. children from the marriage of parents related by blood who are expected to have a larger number of homozygous) than the general population[8]. In this regard, results from the investigation carried out to estimate the level of homozygosity in individuals with different health disorders showed that from a total of 20 recessive characters, 6 of them were found to be more frequent in students with mental retardation (from special schools) compared to control individuals[9].

One of the findings when elaborating the susceptibility of different genotypes relates to carriers of blood group A,

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who are more susceptible to a larger number of malignant tumors, *ulcus duodeni* and *diabetes mellitus*, whereas blood group O is more sensitive to gastric diseases and cholera but it is less prone to syphilis[10], whereas in a number of other diseases, no association was observed[11].

One of the most significant associations with disease susceptibility is that of blood groups A, B and AB (non-group O) with arterial and venous thromboembolism[12]. It has also been reported and argued that rate of infections can be directly linked to ABO phenotype where group O is more sensitive to severe infections from Escherichia coli, while pox is most severely »attacking« group A[10,12]. One of the earliest investigations in this area presented data on the frequency of ABO groups and the Rh factor in 1257 patients who suffered from bronchial carcinoma: individuals with blood group 0 correlated better with undifferentiated tumors than glandular or squamous tumors[13].

The presence and frequency of certain phenotypes can be taken as a basis to determine the genetic predisposition to certain diseases. Features such as ear lobe (free/attached), tongue rolling (roller/nonroller), chin (normal/cleft), hand clasping (right thumb over/left thumb over), handedness (right/left) as well as the blood groups ABO and the Rh systems were therefore examined in patients with lung disease for eventual assocation between the appearance of these phenotypes and lung diseases.

Material and Methods

In this work 2777 patients with lung diseases (mean age 47) and 2778 healthy (control) individuals (mean age 45) from all regions of Kosovo's mainly Albanian nationality were examined for the following morpho- and physiological features (Table 1): ear lobe free – vs. ear lobe attached, (OMIM number 128900); normal chin vs. cleft chin (OMIM number 119000); tongue roller vs non-roller (inability of tongue rolling) (OMIM number 189300); right-handed vs. left-handed (OMIM number 139900, gene location 2p12-q22); hand clasping – right thumb over vs. hand clasping- left thumb over (OMIM number 139800); blood groups: ABO system (OMIM number 110300, gene location 9q34); Rhesus factor (OMIM number 111680, gene location 1p36.2-p34).

TABLE 1
ANALYSED MORPHOLOGICAL TRAITS/PHENOTYPES

Phenotype 1	Phenotype 2
Eear lobe free (ELF)	Ear lobe attached
Normal chin (NC)	Cleft chin*
Tongue roller (TR)	Non-rollers (tongue rolling)
Righthanded (RH)	Lefthanded
Hand classping- Right thumb over (HC)	Hand classping- Left thumb over

* »Cleft Chin« in our investigation refers to typical expression of this phenotype (mostly as the hole in the chin).

Investigations were conducted in the period 2003 – 2010 in regional lung disease clinics of 28 municipalities in Kosovo. The study included patients which were diagnosed with any kind of lung diseases such as bronchopneumonia, acute bronchitis, pneumonia, infiltrative lung diseases, obstructive lung diseases, tuberculosis etc. The control group includes data obtained from blood transfusion patients in the respective hospitals and from healthy persons knowing their blood group and Rh factor. The control individuals were randomly selected in 24 municipalities.

Control individuals and patients were analyzed for the selected phenotypes (discriminating between presence of the phenotypes 1 and 2, blood groups (ABO system) and Rhesus factor (negative vs positive) at 24 (control) and 28 (patients) different municipalities in Kosovo allowing gender matched cohorts of control individuals and patients respectively. The frequencies of alleles for blood groups ABO and the Rh system were calculated[14-16].

Statistical methods

Normal distribution of the data grouped by municipalities was confirmed by applying the »One-Samples« Kolmogorov-Smirnov Test. Student's double sided t-test for independent samples was performed to determine the level of significance for comparing the means of the patients and control group. »One-way« ANOVA, Brown-Forsyth and Welch-test followed by Tamhane-T2 *post-hoc* testing was used to examine the degree of significance for gender related differences between the patients and the control group.

Results

A study overview including frequencies of the different phenotypes in controls and patients suffering from lung diseases, blood groups and Rhesus factor positives in controls and patients suffering from lung diseases and municipality based assignement is given in the Tables 2, 3 and 4.

Analysis for differences between the control (N=2777) and the patient (N=2778) group of the primary scores (not assigned to municipality groups) using the Mann-Whitney U Test revealed significant differences ($p<0.05$) for ELF, NC, TR, blood groups A and AB and Rh+. In order to employ normal distribution based statistics (ANOVA, t-test), the scores from the different municipalities were assigned as individual samples (N=24 for control and N=28 for patients). Data distribution analysis by Box-plots (Figure 1) indicates that data distribution is homogeneous in most locations with moderate skewness. Most of the outliers are „mild" and only two datapoints have been identified as strong (extreme) outliers: Sharr, showing the highest score of control individuals with blood group A and Shterpca revealing the highest score of control individuals with blood

TABLE 2
STUDY OVERVIEW – FREQUENCIES OF THE DIFFERENT PHENOTYPES IN CONTROLS AND PATIENTS SUFFERING FROM LUNG DISEASE

Control Group[a]															
	ear lobe free (ELF)			normal chin (NC)			tongue roller (TR)			Righthand (RH)			right clasper (RC)		
	0	1	Total	0	1	Total	0	1	Total	0	1	Total	0	1	Total
N	1238	959	2197	1251	986	2237	1037	778	1815	1463	1181	2644	973	740	1713
Total	1552	1226	2778	1552	1226	2778	1552	1226	2778	1552	1226	2778	1552	1226	2778
%	,798	,782	,791	,806	,804	,805	,668	,635	,653	,943	,963	,952	,627	,604	,617
Patient Group[a]															
	ear lobe free			normal chin			tongue roller			righthand			right clasper		
	0	1	Total	0	1	Total	0	1	Total	0	1	Total	0	1	Total
N	1086	910	1996	1113	939	2052	799	640	1439	1435	1192	2627	955	761	1716
Total	1520	1257	2777	1520	1257	2777	1520	1257	2777	1520	1257	2777	1520	1257	2777
%	,714	,724	,719	,732	,747	,739	,526	,509	,518	,944	,948	,946	,628	,605	,618
a.0 – female, 1 – male															

TABLE 3
STUDY OVERVIEW – BLOOD GROUPS AND RHESUS FACTOR POSITIVES IN CONTROLS AND PATIENTS SUFFERING FROM LUNG DISEASE

Control Group[a]															
	BG_A			BG_B			BG_AB			BG_O			Rh		
	0	1	Total	0	1	Total	0	1	Total	0	1	Total	0	1	Total
N	573	429	1002	232	214	446	93	97	190	654	486	1140	1288	1049	2337
Total	1556	1226	2782	1556	1226	2782	1556	1226	2782	1556	1226	2782	1552	1226	2778
%	,368	,350	,360	,149	,175	,160	,060	,079	,068	,420	,396	,410	,830	,856	,841
Patient Group[a]															
	BG_A			BG_B			BG_AB			BG_O			Rh		
	0	1	Total	0	1	Total	0	1	Total	0	1	Total	0	1	Total
N	605	493	1098	206	189	395	66	54	120	642	521	1163	1216	1007	2223
Total	1520	1257	2777	1520	1257	2777	1520	1257	2777	1520	1257	2777	1520	1257	2777
%	,398	,392	,395	,136	,150	,142	,043	,043	,043	,422	,414	,419	,800	,801	,801
a.0 – female, 1 – male															

TABLE 4
STUDY OVERVIEW –MUNICIPALITY BASED ASSIGNMENT

Report											
group		ELF	NC	TR	RH	RC	Rh+	BG_A	BG_B	BG_AB	BG_O
Control (24)	Mean± Std. Dev.	73.97±7.74	79,64±5.93	67.76±10.09	9217± 6.13	66,92±13.20	82.04±9.97	41.27±17.05	14.94±7.53	7.84±8.41	35.93±10.86
Lung disease (28)	Mean± Std. Dev.	69.58±10.88	73.28±12.02	54.19±10.37	94.70±3.78	63.27±7.26	79.60±8.19	37.50±6.73	14.83±5.88	3.72±3.35	43.91±8.95

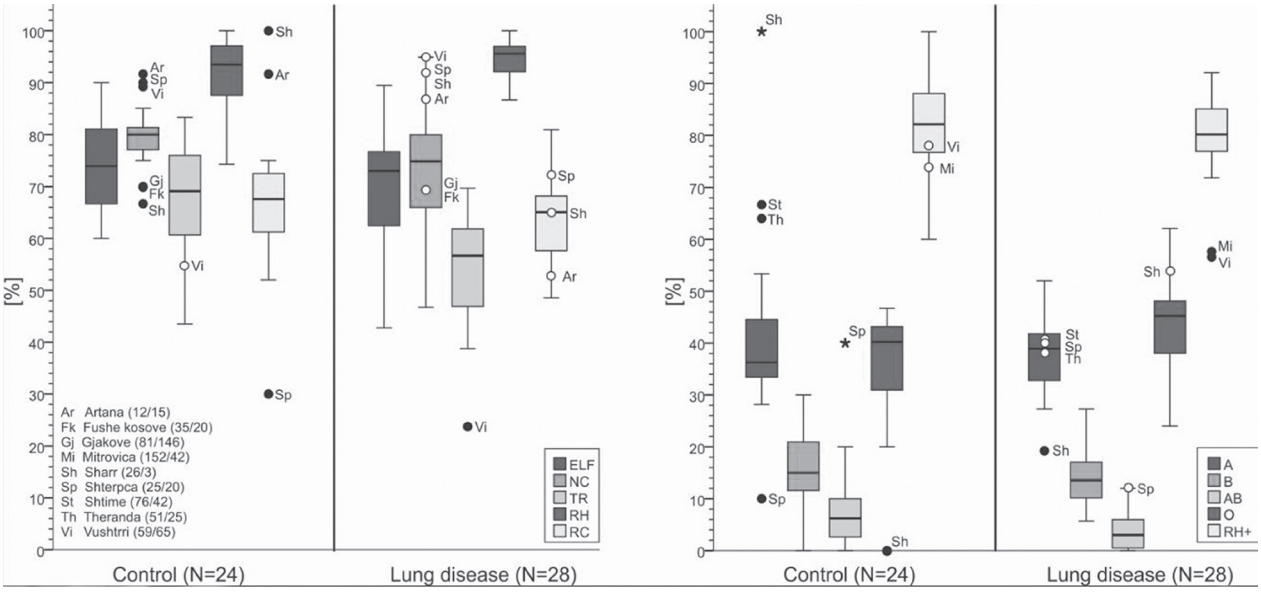

Fig. 1. Frequencies of the different phenotypes, blood-group distribution and presence of the rhesus-factor. Box-plot analysis shows the distribution of the different parameters at the investigated municipalities. Boxes represent the 25% (lower border), 50% (line – median) and 75% (upper border) percentille, whiskers the statistically valid lowest and highest datapoints. Closed dots refer to cases which have been identified as mild outliers (1.5 – 3 fold of the interquartille range), asterisks identify statistically exctreme outliers (> 3 fold of the interquartille range). Municipality abbreviations are given in the left panel; numbers in brackets refer to the numbers of scored controls/ patients.

group AB. Since the numbers of individuals scored in these locations are small compared to the whole number of individuals scored (Sharr 0.3% of all control individuals of blood group A and Shterpca 4.2% of all controls blood group AB) these cases have not been excluded from further statistical analysis, the data still being normally distributed.

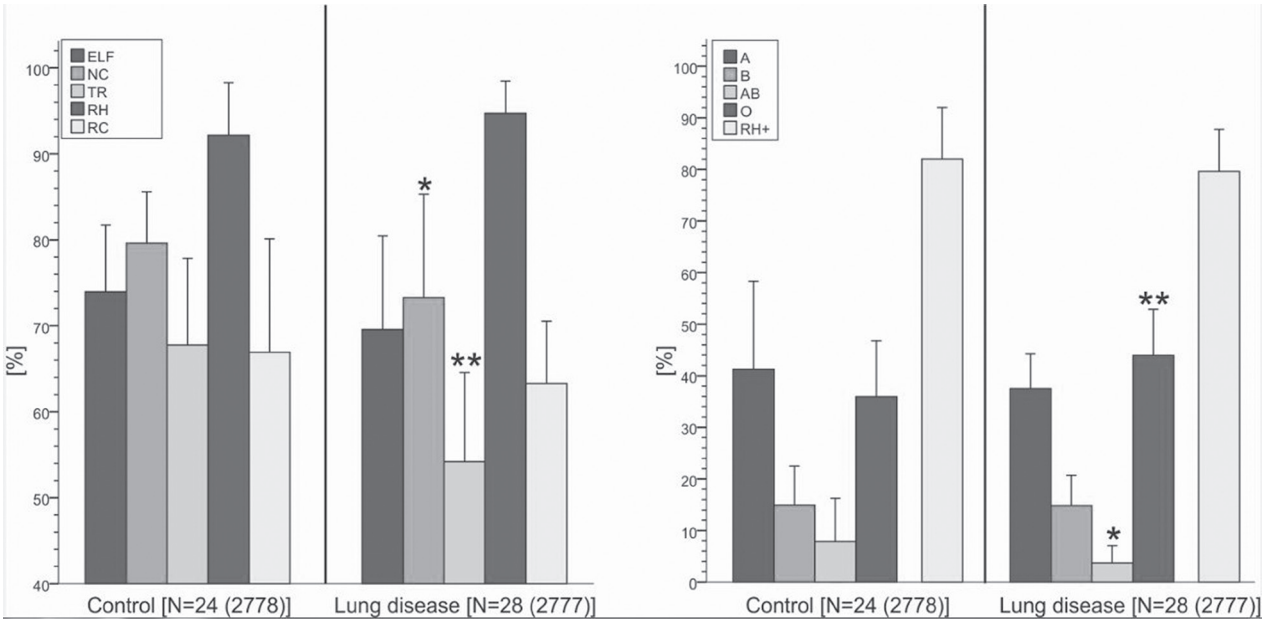

*Fig. 2. Influence of disease state on the occurrence of the different phenotypes and blood parameters. Bars represent the mean ± SD of the percentages of individuals showing the indicated phenotypes and blood specificities. One-way ANOVA (ANOVA, Brown-Forsyth test, Welch-test) proofs significant ($P < 0.05$) differences between controls and lung disease patients for NC, TR, blood groups AB and 0. * $P < 0.05$, ** $P < 0.005$ refers to pairwise comparisons (control vs. patient) by the Student's double-sided t-test for independent samples. N refers to the different sampling municipalities, the total number of scored individuals given in parentheses.*

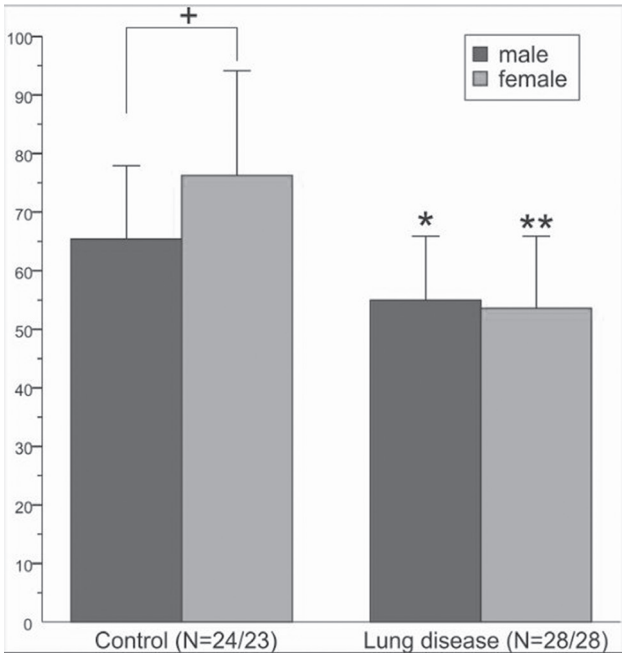

*Fig. 3. Gender dependent differences in tongue rollers. Bars represent the mean ± SD of the percentages of individuals with tongue rolling ability. One-way ANOVA (ANOVA, Brown-Forsyth test, Welch-test) proofs significant (P < 0.05) differences between controls and lung disease patients. * P < 0.05, ** P < 0.005 compared to the corresponding controls (ANOVA post-hoc testing, Tamhane T2); Note the significant difference (+ P < 0.05; One-way ANOVA) between male and female control individuals.*

Notably, for NC, the median (50 percentile) and in particular the 25 percentile is lower in lung disease which results in a significant difference between control and patients in the t-test (as shown in Figure 2). However, detailed analysis of the NC phenotype shows that there are no differences between controls and lung disease in Gjakova(Gj) and Fushe Kosova(Fk) and that there are moderate differences in Vushtrri (Vi) and Shterpce (Sp), while the incidence of the NC phenotype is approximately 12% higher among lung disease patients as compared to the control in Shterpce (Sh). For TR, box-plot analysis apparently indicates a less conflicting situation. Indeed, this effect of lung disease is strongly supported by the findings obtained in Vushtrri (Vi) where a value between the 25 percentile and < 1.5 times the interquartile range becomes an outlier (lower range!) in the patients group.

Regarding blood group A, the opposite scenario is found: Here the controls show a higher variance compared to the patients due to »severe« Shterpce (Sp) and Sharr (Sh) and »moderate« Shtime (St) and Therande (Th) outliers. With the exeption of Shterpce (Sp), blood group A is less frequent in lung disease patients at these sites.

Although this finding is most pronounced in Sharr (Sh), where all controls (N=26) had blood group A, but only 19% of the patients, the low number of investigated patients (N=3) complicates statistical interpretation. In contrast to NC, there is no significant difference between controls and patients with respect to blood group A. Thus, such differences may be related to random rather than disease related effects.

t-Statistics, ANOVA

ANOVA and t-Statistics indicate, that the NC and TR phenotype is significantly less frequently seen in lung patients (Figure 2). Regarding blood groups, AB is significantly less frequent ($p \leq 0.05$) but blood group 0 is more often ($p \leq 0.005$) found in lung patients. Statistical analysis also revealed a gender specific difference for tongue rollers in the population examined (Figure 3). In the control group there are significantly more ($p < 0.05$) female tongue rollers than male, while this difference disappears in the lung disease group. These results also indicate that tongue rollers of both sexes have a significantly lower risk of developing lung disease (males: $p < 0.05$; females: $p < 0.005$).

Alele frequencies of the ABO and Rh blood group systems

Based on the data obtained from the examination of individuals in the control group, allele frequencies for the ABO and Rh system for the Kosovar population were calculated. The frequencies of alleles of the ABO system were: $I^O = 0.64$; $I^A = 0.24$ and $I^B = 0.12$, whereas, alleles D and d of the Rh locus gave frequencies of: D = 0.60 and d = 0.40.

Discussion

The analysis of the genetic structure of populations is among the most important approaches to investigate the changes that occur in human populations with special interest in genetic risk assessment and prediction of inheritance and predisposition of certain diseases[1,2]. In this context, the relative importance of natural selection in the distribution of blood groups in humans has been the subject of discussion and investigation since the last century when it was noticed that the frequencies of blood groups vary among different populations and confer varying degrees of susceptibility to various infectious diseases as Helicobacter pylori, cholera infections etc.[12]. In one of the classical papers in the field[17], it was presented a higher incidence of some genetic diseases in certain ethnic groups and explained that the high frequency of some recessive lethal alleles are favored by selection for other reasons, either because of the linkage with other genes or because the heterozygous state was favorable to the carrier. A study on patients suffering from tuberculosis, and representing three ethnical groups (Chinese, Malayzians and Indians), revealed that Chinese blood group O carriers were more resistant to infection than others[18]. Given the nature of recessive mutations, the high degree of homozygosity is reported to accompany a range of diseases and it can be assumed that the genetic basis for a large number

of diseases involves recessive alleles. The extent of genetic load thereby will depend on the specific population groups inhabiting the same place and on genetic adaptations of these population groups under particular environmental conditions[4,19-25]. Based on research conducted in isolated genetic populations, it turned out that inbreeding is significant predictor for a number of diseases as coronary heart disease, stroke, cancer, uni/bipolar depression, asthma, gout, and peptic ulcer[26].

Regarding the mode of inheritance for selected traits in our investigation such as ear lobe, tongue rolling, blood groups from ABO and Rh system, there is »agreement« among researchers concerning dominant and recessive phenotypes, whereas for features such as cleft chin, handedness and hand clasping, authors differ in their conclusions. Thus, the presence of the cleft chin described by some authors as a recessive phenotype[27] in »Online Mendelian Inheritance in Man«[5], this feature appears as dominant. Concerning hand clasping, there is also »disagreement« in data from the literature starting from the conclusion that hand clasping is: a) not determined by genetic factors[28] b) may be under genetic control[29] or c) that it is under polygenic influence[30]. According to OMIM[5], right-handedness is an incomplete dominant phenotype and takes into account the possibility of multifactorial influence[31], while others[32] conclude that there is no convincing data that genes play a role in handedness. Therefore, this feature is also analyzed classifying phenotypes as R (right) and L (left).

Our results show that the NC and TR phenotype is significantly less frequently seen in lung patients. Regarding blood groups, AB was found to be significantly less frequent whereas blood group O is more often found in patients suffering from lung diseases, although only 6.8% and 4.3% of the control and patient group have blood group AB, eventually influencing statistical significance. Case-analysis indicates a potentially relevant aspect of the study: outliers contribute markedly more to the overall frequency of phenotypes in the control group compared to the patients group, especially in the case of NC and blood group A. On the other hand, variances are higher in NC (patient) and blood group A (control). For NC this indicates that NC controls are more homogeneous and »extreme« values rather become outliers, since they are out of the interquartile range. In contrast, in lung disease patients the NC phenotype generally shows a higher variation as evident from the interquartile range also including values which would represent outliers in the control group.

Regarding gender based statistics, lung disease apparently abolishes a sex-related difference in tongue rollers as indicated by ANOVA – post hoc testing and opens a field of discussion on X-linked phenomena.

Last, it is worth to mention that the lung disease issue is very complex taking into consideration that several genes are likely to be implicated in the pathogenesis for chronic obstructive pulmonary disease[33] whereas more than 300 genes appear to be potentially involved in the development and expression of only one of the lung diseases – asthma[34]. In addition, it has been reported that increased homozygosity and accumulated genetic loads are not the sole cause of allergic bronchial asthma indicating the importance of polygenic regulation[35,36]. Nevertheless, the findings from this study demonstrate that an increased recessive homozygosity in patients (of some of the features analysed), as well as the specific associations obtained, may contribute to a predisposition for lung diseases in general. In a further step the underlying database will be increased with additional cases and controls to allow statistical evaluation of specific lung diseases and the determination of confounding variables.

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DISTRIBUCIJA UČESTALOSTI I VEZA IZMEĐU NEKIH MORFOLOŠKIH I FIZIOLOŠKIH SVOJSTAVA KOD BOLESNIKA S PLUĆNIM BOLESTIMA NA KOSOVU

S A Ž E T A K

Cilj ovog rada bio je istražiti raspodjelu pojedinih fenotipova kod pacijenata s bolestima pluća kao i njihove eventualne povezanosti s rizikom razvoja bolesti pluća. U tu svrhu je ispitano 2777 pacijenata s bolestima pluća i 2778 zdravih ispitanika iz cijelg područja Kosova za pojavnost sljedećih odabranih fenotipova: ušna resicu odvojena (ELF)/ušna resica spojena, normalna brada (NC)/rascjep brade, jezik koji može u valjak (TR)/ne može u valjak, preklop desnog palca tijekom stiskanja ruke(HC)/preklop lijevog palca tijekom stiskanja ruke, dešnjaci (RH)/ljevaci. Osim toga su promatrani I uzorci krvi iz grupa ABO sustava i prisutnost ili odsutnost Rhesus faktora kao uočenih fenotipskih markera. Dobiveni rezultati pokazuju značajne razlike između kontrolne skupine i bolesnika s plućnim bolestima za NC ($p \leq 0.05$) i TR ($p \leq 0.005$), kao i za krvne grupe AB ($p \leq 0.05$) i O ($p \leq 0.005$). Ovi rezultati na kraju ukazuju na povećane razine genetskog opterećenja kao rezultat povećanog broja homozigota na nekim lokacijama gena. To uzrokuje povećanu učestalost nekih recesivnih fenotipova u pacijenata s bolestima pluća. Zajedno sa specifičnim primjećenim vezama, ovi preliminarni rezultati mogli bi poslužiti kao osnova za daljnja dubinska istraživanja s obzirom na vrste bolesti pluća, profesionalne izloženosti i prehrambenim navikama, a time i očekuje se da će doprinijeti razumijevanju predispozicija i osjetljivosti na plućne bolesti.