

The Familial Occurrence of Obstructive Sleep Apnoea Syndrome (OSAS)

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Abstract: The aim of the study was to compare the incidence of obstructive sleep apnoea/hypopnoea syndrome (OSAS) in relatives of subjects with OSAS and in relatives without OSAS but with clinical symptoms of this disease. The study group consisted of 186 relatives of patients with OSAS and 117 relatives of patients with symptoms of OSAS in whom the disease was not confirmed by polysomnography. They were all mailed a questionnaire with questions concerning anthropometric data, the presence of symptoms typical for OSAS and the presence of concomitant diseases. Analysis of the obtained data revealed an increased frequency of snoring, sleep apnea and nycturia in the relatives of patients with OSAS when compared to relatives of patients without OSAS, but the difference was not statistically significant. The incidence of daytime OSAS symptoms was significantly higher in the group of relatives of patients with OSAS. No differences in the incidence of arterial hypertension, ischemic heart disease and diabetes mellitus were found.

In order to confirm the presence of OSAS in family members with typical symptoms, we performed polysomnography. Were diagnosed OSAS in 20 (29.4 %) relatives of patients with OSAS and only in 8 (18.2) relatives of patients without OSAS.

These results confirm the increased incidence of OSAS in patients with a family history of this disease.

Keywords: OSAS, familial occurrence.

INTRODUCTION

Obstructive sleep apnoea syndrome (OSAS) can be defined as the coexistence of excessive daytime sleepiness with irregular breathing at night. OSAS is common in the population and affects approximately, 9% men and 2.5% women in middle age in Poland [1]. Many studies have been performed in order to identify risk factors for OSAS. Familial occurrence and genetic factors have also been investigated.

In 1978 Strohl *et al.* described a family with OSAS [2]. In 1993 Douglas *et al.* found familial clustering of symptoms typical for OSAS such as snoring and apnoeas [3]. Familial aggregation was shown in Susan Redline's study and this study has been the basis of many genetic investigations of OSAS [4]. To the author's knowledge, familial occurrence of OSAS in the Polish population has not been investigated. The aim of the study was to compare the incidence of OSAS in relatives of subjects with OSAS and in relatives without OSAS but with clinical symptoms of this disease.

MATERIAL AND METHODS

We randomly selected 50 patients with OSAS and 50 snoring patients who had all undergone overnight polysomnography. The polysomnography involved the

EEG, EOG and EMG records allowing to determine the sleep structure and a number of wakeups. A kind of breathing disorders during sleep was recorded thanks to the continuous airflow through the respiratory tracts (measurement with the use of a thermistor), chest and stomach movements (with the use of two piezoelectric belts) and the assessment of the oxygen saturation of arterial blood with the use of a pulse oximeter. The diagnosis of OSAS was established when the AHI (apnoea/hypoapnoea index) value exceeded 5 and over 10 points in the Epworth Sleepiness Scale (ESS). We then mailed our questionnaires to all first degree relatives of the index patients in each group.

Both groups consisted of first-degree relatives (parents, siblings and children) of the above mentioned patients. They were all sent questionnaires enquiring about age, sex, obesity and symptoms associated with OSAS (snoring, daytime sleepiness and apnoeas). Two types of questionnaires were used. The full-length questionnaire (Figure 1) contained detailed questions on the incidence, frequency and intensity of typical symptoms for OSAS. The mini-questionnaire (Figure 2) contained questions about the occurrence and intensity of the symptoms and it was filled by the family of the included first degree relative with which direct contact was impossible.

We sent questionnaires to 503 family members of both groups (301 questionnaires from the FI group and 202 questionnaires from the FH group). The subjects in

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Name:..... Male/Female:..... Familial relationship.....
 Height:..... Weight:..... Age:.....

1. How often do you snore?
 - a. Nearly every day
 - b. 3-4 times a week
 - c. 1-2 times a week
 - d. 1-2 times a month
 - e. never or nearly never
2. Has anyone noticed that you stop breathing during your sleep?
 - a. Nearly every day
 - b. 3-4 times a week
 - c. 1-2 times a week
 - d. 1-2 times a month
 - e. never or nearly never
3. How often do you sweat intensively during sleep?
 - a. Nearly every day
 - b. 3-4 times a week
 - c. 1-2 times a week
 - d. 1-2 times a month
 - e. never or nearly never
4. How often do you get up to the toilet during the night?
 - a. Nearly every day
 - b. 3-4 times a week
 - c. 1-2 times a week
 - d. 1-2 times a month
 - e. never or nearly never
5. How often do you feel tired or fatigued after your sleep?
 - a. Nearly every day
 - b. 3-4 times a week
 - c. 1-2 times a week
 - d. 1-2 times a month
 - e. never or nearly never
6. How often do you feel tired/fatigued after waking up?
 - a. Nearly every day
 - b. 3-4 times a week
 - c. 1-2 times a week
 - d. 1-2 times a month
 - e. never or nearly never
7. Have you ever fallen asleep while driving a vehicle?
 - a. YES
 - b. NO
8. Do you have high blood pressure?
 - a. YES
 - b. NO
9. Do you have a ischaemic heart disease?
 - a. YES
 - b. NO
10. Do you have a diabetes?
 - a. YES
 - b. NO

Figure 1: Full-length questionnaire.

Name:..... Male/Female:..... Familial relationship.....
 Height:..... Weight:..... Age:.....

Snoring:	YES	NO
Stop breathing during sleep:	YES	NO
Sleepiness during day:	YES	NO
Hypertension:	YES	NO
Heart disease:	YES	NO
Diabetes:	YES	NO

Figure 2: Mini-questionnaire.

both groups had a similar age (55.0 ± 18.9 in group FI vs. 54.1 ± 19.2 in group FH) and body mass index (27.6 ± 8.5 in group FI vs. 27.4 ± 6.4 in group FH). After receiving the answers, we selected subjects with symptoms typical for OSAS (47 subjects from group FI and 16 subjects from group FH) and referred them to

polysomnography in order to confirm or exclude the presence of OSAS.

The algorithm of the patient selection and methods applied are presented on Figure 3. The comparison of the data obtained from the questionnaires and in the

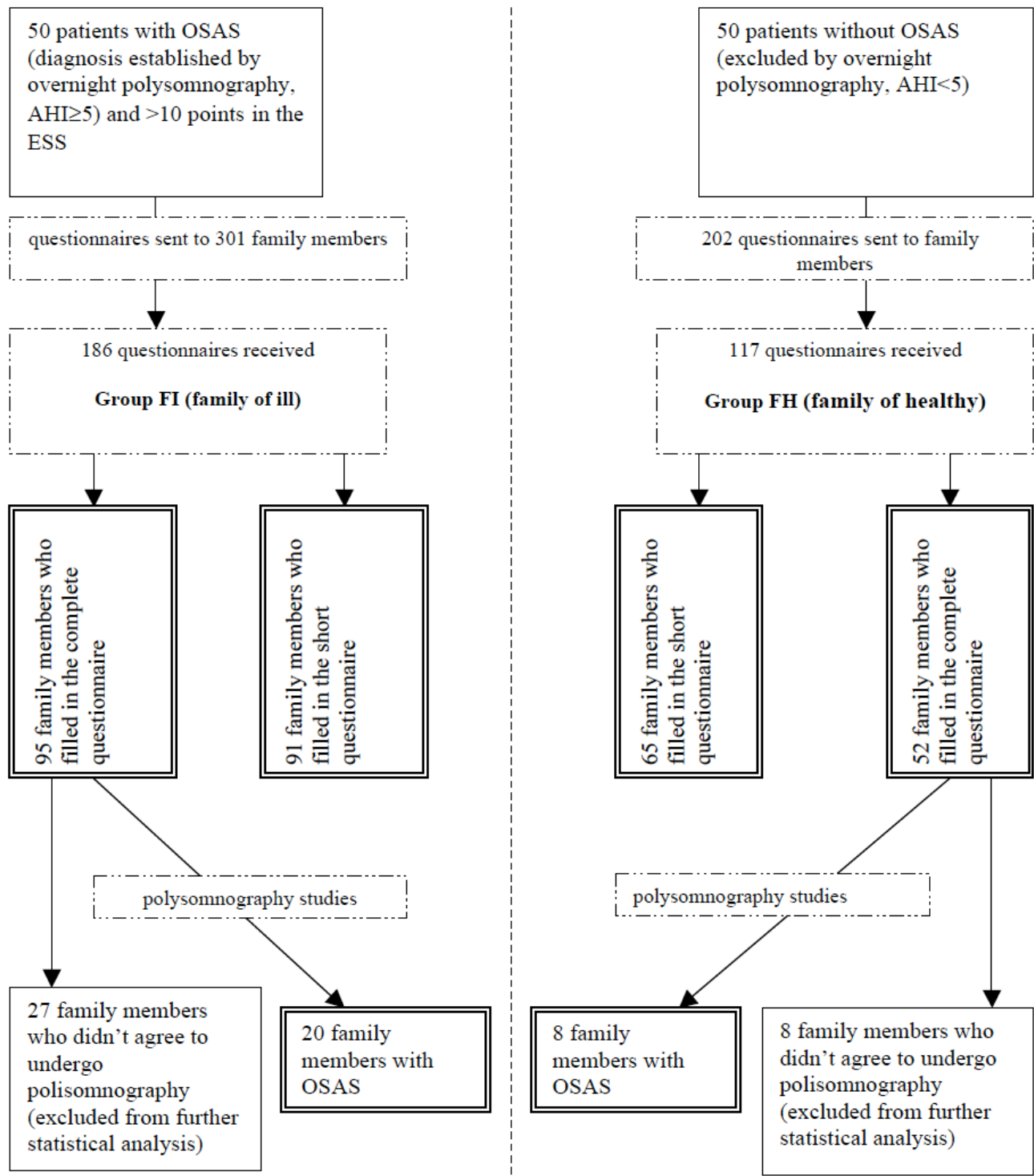


Figure 3: Algorithm demonstrating the patient selection and methods.

results polysomnography from both groups are shown in Table 1 and 2.

Data are presented as average ± standard deviation (SD). Non-parametric tests and logarithmic transformations were applied in the analysis of nonnormally distributed variables.

RESULTS

We received 303 questionnaires from the total of 503 sent (60.2%): 186 questionnaires were from the FI group and 117 questionnaires from the FH group (61.8 % vs. 57.8 %, difference not significant). Analysis of the data revealed an increased frequency of snoring, sleep apnea and nocturia in the relatives of patients with

Table 1: Comparison of the Data Obtained from the Questionnaires from Group FI and FH

	Group FI	Group FH	P
<i>Age(yrs)</i>	55.0±18.9	54.1±19.2	ns
<i>Body mass index (kg/m²)</i>	27.6±8.5	27.4±6.4	ns
<i>Snoring</i>	61 (52.1 %)	107 (57.5 %)	ns
<i>Everyday snoring</i>	28 (56.0 %)	63 (65.6 %)	p<0.01
<i>Apnoea</i>	31 (26.5 %)	65 (35.0 %)	ns
<i>Perspiration</i>	23 (43.4 %)	54 (56.3 %)	ns
<i>Nycturia</i>	33 (62.3 %)	66 (69.5 %)	ns
<i>Somnolence after waking up</i>	30 (56.6 %)	80 (83.3 %)	p<0.001
<i>Involuntary daytime sleep</i>	14 (25.0 %)	43 (44.8 %)	p<0.05
<i>Sleepiness during car driving</i>	3 (7.7 %)	18 (24.7 %)	p<0.05
<i>Arterial hypertension</i>	13 (23.2 %)	22 (22.9 %)	ns
<i>Ischemic heart disease</i>	6 (10.7 %)	15 (15.6 %)	ns
<i>Diabetes</i>	4 (7.1 %)	9 (9.4 %)	ns

OSAS when compared to relatives of patients without OSAS, but the difference was not statistically significant. The incidence of daytime OSAS symptoms was significantly higher in the group of relatives of patients with OSAS. We did not find differences in the incidence of arterial hypertension, ischemic heart disease and diabetes mellitus in both groups. These results are presented in Table 1.

Polysomnography revealed a higher value of AHI and lower mean oxygen saturation (SaO₂) during sleep in subjects from the FI group. Moreover, the incidence of OSAS (i.e. AHI ≥ 10) was significantly higher in the FI group when compared to the FH group (29.4 % vs. 18.2%, p < 0.05). The comparison of the results of polysomnography is presented in Table 2.

Table 2: Comparison of the Results of Polysomnography in Groups FI and FH

	Group FI	Group FH	P
AI (apnoea index)	15.2±26.7	3.6±9.8	p<0.05
HI (hypopnoea index)	6.5±9.3	5.3±5.9	ns
AHI (apnoea/hypopnoea index)	22.8±28.4	8.8±11.2	p<0.05
mean SaO ₂	92.8±3.5	93.9±2.0	p<0.05
min. SaO ₂	79.5±12.0	85.6±5.2	p<0.01
family members with OSAS (%)	20 (29.4)	8 (18.2)	p<0.05

DISCUSSION

The first well-documented case of familial obstructive sleep apnoea syndrome was reported by

Strohl *et al.* in 1978. [2] There is now firm evidence that OSAS is more common in the family members of OSAS patients than in the general population [4]. This relationship is independent of the well-known familial nature of obesity [5]. As factors under investigation may be genetically determined, many authors suggest possible contributors: familial facial structure, differences in selective deposition of adipose tissue around the upper airway, upper airway dilator muscle function, ventilatory control and susceptibility to sleepiness [5, 6]. However, the precise mechanism of familial association of OSAS requires further research.

The aim of the study was to compare the incidence of OSAS in relatives of subjects with OSAS and in relatives without OSAS but with clinical symptoms of this disease. The study reported concerned only first-degree relatives. We have proved an increased frequency of snoring and the incidence of daytime OSAS symptoms in the relatives of patients with OSAS when compared to relatives of patients without OSAS. These results confirm the observations of Guilleminault [7] and Redline [8].

Analysis of the polysomnography studies revealed an increased value AHI and decreased mean SaO₂ during sleep in relatives of patients with OSAS. This is in accordance with previous studies [4, 7]. The study in Iceland a different approach – a genealogy strategy. In this study first-degree relatives of patients with OSAS had an about 2-fold increased risk of OSAS [9]. Moreover, we have diagnosed OSAS in 20 (29.4 %) relatives of patients with OSAS and only in 8 (18.2%)

relatives of patient without OSAS. These data confirm a significant familial aggregation of OSAS that appears to be independent of familial similarities in weight and in snoring.

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Received on 25-02-2014

Accepted on 06-03-2014

Published on 02-04-2014

<https://doi.org/10.12974/2312-5470.2014.01.01.4>

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