Otology

Prevalence of prelingual deafness in Italy

La prevalenza della sordità prelinguale in Italia

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SUMMARY

Neonatal hearing loss is the most frequent sensorial congenital defect in newborns. No data are available on worldwide prevalence of congenital deafness. World Health Organization (WHO) data indicate 1-4 cases per 1,000 individuals, with a considerable increase in developing countries. A prevalence exceeding 1 per 1,000 however, indicates a serious public health problem calling for urgent attention. Aim of the study was the evaluate the prevalence of prelingual deafness in the Italian population and determine the socio-demographic characteristics of the condition. Data were provided by the National Institute of Social Insurance (INPS) and the Italian Central Statistics Institute (ISTAT) and were collected in 18 out of the 20 Italian regions (98.2% of total population). All subjects recognized as deaf-mute by a special medical committee were included. According to law No. 509/1988, they had to present a mean bilateral sensorineural-hearing impairment, detected in neonatal age, which caused the damage in speech development and equal to 60 dB or more for 500-, 1,000- and 2,000-Hz frequency tones in the better ear. Prevalence rates were calculated according to region and age bracket using updated population data from census 2001. Statistical analyses were performed using the SPSS statistical software package. A total of 40,887 cases of prelingual profound sensorineural hearing loss ≥ 60 dB were detected in Italy in 2003, for a total prevalence rate of 0.72 per 1,000. The hearing impairment prevalence differs according to sex. The overall prevalence is 0.78 per 1,000 for males and 0.69 per 1,000 for females ($p < 0.001$). The hearing impairment prevalence differs according to region of residence ($p < 0.001$). The geographic distribution of prelingual deafness was found to be: North 15,644 cases (0.63 per 1,000), Central Italy 7,111 cases (0.64 per 1,000), South and Islands 18,132 (0.87 per 1,000). The prelingual hearing loss is highly prevalent in South Italy (Basilicata, Calabria and Sicily). For the southern regions of Italy, the rate observed in the 50-64 and > 64 age groups reached 1.27 and 1.15, respectively. This phenomenon may have been due, in part, to the epidemic incidence of maternal rubella which occurred in the 40's and 50's (in Italy, the rubella vaccination was only recommended starting from 1972), and, in part, to the habit of contracting consanguineous marriages. Data from the Vatican Archives on 520,492 consanguineous marriages, for which dispensation was requested in the period 1911-1964, indicate that in the years 1935-1939, in small villages in South Italy (Basilicata, Calabria, Sicily) consanguineous marriages accounted for over 40% of marriages.

KEY WORDS: Sensorineural hearing loss • Congenital hearing loss • Prelingual deafness • Prevalence

RIASSUNTO

La sordità neonatale risulta essere il difetto congenito ereditario più frequente nei neonati. I dati riguardanti la prevalenza della sordità congenita nella maggior parte dei Paesi non sono disponibili. Il World Health Organization (WHO) fornisce una stima presunta di prevalenza nella popolazione tra 1-4 casi su 1.000 abitanti, con un aumento sensibile nei soggetti svantaggiati e nei Paesi in via di sviluppo. Una prevalenza superiore a 1 caso su 1.000 rappresenta un grave problema di sanità pubblica che necessita di attenzione urgente. Il presente studio ha come obiettivo quello di stimare la prevalenza della sordità prelinguale nella popolazione italiana, descriverne le caratteristiche socio demografiche e la distribuzione geografica per regione. I dati provengono dalla banca dati dell'Istituto Nazionale di Previdenza Sociale (INPS) e dall'Istituto Nazionale di Statistica (ISTAT) e sono stati raccolti in 18 su 20 regioni italiane (98,2% della popolazione italiana). Sono stati arruolati allo studio tutti i casi di sordità prelinguale riconosciuti come sordomuti da una speciale commissione medica. I soggetti sulla base della legge n. 509/1988, dovevano presentare una perdita della capacità uditiva media di tipo neuro-sensoriale bilaterale ≥ 60 dB per le frequenze 500-1.000-2.000 Hz nell'orecchio migliore, avvenuta in epoca neonatale e tale da alterare lo sviluppo del linguaggio. Sono stati calcolati i tassi di prevalenza per classi di età e per regione utilizzando i dati aggiornati al 2003 dell'ultimo censimento nazionale della popolazione relativo al 2001. L'elaborazione statistica è stata eseguita utilizzando il pacchetto statistico SPSS. I casi con perdita della capacità uditiva neuro-sensoriale prelinguale ≥ 60 dB presenti in Italia nel 2003 sono stati 40.887 (20.915 maschi e 19.778 femmine), con una prevalenza nella popolazione di 0,72 per 1.000 abitanti (popolazione italiana = 56.780.305). La prevalenza della sordità differisce nei due sessi: 0,78 per 1.000 nei maschi e 0,69 per 1.000 nelle femmine ($p < 0,001$). Una differenza significativa ($p < 0,001$) dei tassi di prevalenza è presente nella distribuzione geografica dei casi di sordità prelinguale: sono risultati 15.644 i casi nel Nord (0,63 per 1.000), 7.111 nel Centro (0,64 per 1.000) e 18.132 nel Sud ed Isole (0,87 per 1.000). La perdita della capacità uditiva prelinguale è altamente prevalente nell'Italia meridionale ed in particolare in alcune Regioni del Sud e delle Isole. In particolare in Basilicata, Calabria e Sicilia il tasso osservato nelle classi d'età 50-64 e > 64 era rispettivamente di 1,27 e 1,15. Questo fenomeno può essere dovuto in parte all'incidenza epidemica della rosolia materna avvenuta negli anni '40-'50 (in Italia la vaccinazione contro la rosolia è stata suggerita soltanto a partire dal 1972) ed in parte all'abitudine alle unioni tra consanguinei. Dai dati provenienti dagli archivi del Vaticano su 520.492 unioni tra consanguinei per cui doveva essere richiesta la dispensa tra il 1911-1964, un recente studio indica che, negli anni '35-'39, nei paesi di alcune Regioni italiane del Sud (Basilicata, Calabria, Sicilia) i matrimoni tra consanguinei ammontavano ad oltre il 40% delle unioni.

PAROLE CHIAVE: *Ipoacusia neuro-sensoriale • Ipoacusia congenita • Sordità prelinguale • Prevalenza*

Introduction

Neonatal prelingual non-syndromic deafness is the hereditary sensorial defect most commonly observed in newborns, with an estimated incidence of approximately 200/100000 babies born alive, as compared to other congenital disorders which, on newborn screening, have been found to have an estimated incidence of 13/100000 for haemoglobinopathies, 10/100000 for phenylketonuria and 25/100000 for congenital hypothyroidism [1] (Fig. 1).

Data concerning prevalence of congenital deafness are not available for most Countries.

The prevalence estimated in the population, by the World Health Organization (WHO), suggests 1-4 cases per 1,000 inhabitants, with a considerable increase in disadvantaged subjects and developing Countries. A prevalence over 1/1000, however, is considered, by the WHO, to be a serious public health problem requiring urgent attention [2].

According to more recent epidemiological data, in the United States of America, the estimated prevalence of profound deafness observed upon newborn screening is 1-2 per 1000 [3-5].

A large number of studies in samples of Italian newborns, subjected to neonatal screening according to heterogeneous inclusion criteria, show a highly variable prevalence ranging from 0.8 to 2.1 per 1,000 [6-10].

Studies in babies assisted in neonatal intensive care units (NICUs), show that deafness has increased to 4% of all subjects [11-14].

Most of the epidemiological investigations carried out on deafness in small children involved mainly restricted samples of selected populations, and variability in data is the result of heterogeneous samples and different selecting methods.

So far, considering the very small number of newborns undergoing neonatal hearing screening, which currently covers 29.3% of the babies [15] born in Italy, i.e., a total of 156,048 newborns in 2003, the prevalence of the disease cannot be correctly estimated yet, particularly considering the heterogeneous screening is performed in the territory.

It is currently held that in order to better assess the true problem, it is necessary to establish the current prevalence rate of the disease.

Aim of the study was to estimate the prevalence of prelingual deafness ≥ 60 dB in the Italian population and to outline the sociodemographic characteristics and geographic distribution of the disorder.

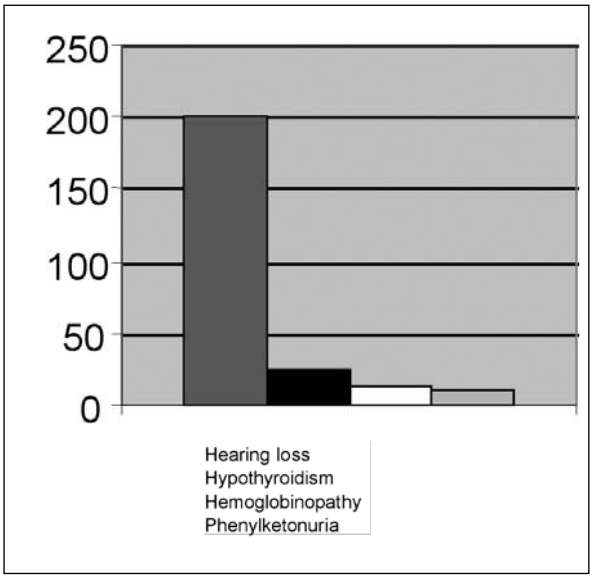

Fig. 1. Incidence of certain congenital diseases per 100,000 newborns.

Material and Methods

Data used in this study were made available by courtesy of the Italian National Institute of Social Insurance (INPS) and the Italian Central Statistics Institute (ISTAT) data banks. As, in Italy, sickness benefits are granted to recognized cases, INPS keeps a record of all cases of prelingual deafness. The data available refer to 18 of the 20 Italian regions (98.2% of the Italian population). All the subjects with prelingual deafness, recognized in 2003 as "deaf-mutes" by a special medical board according to law No. 509, art. 4, dated Nov. 21 1988 [16], were enrolled in the study. Since 1992, in accordance with D.M. (Ministry Decree) No. 293/89 [17] and D.M. No. 148/92 [18], the Ministry of Economy have checked further reliability of disability through a Supreme Medical Committee. Study inclusion criteria comprised: documented clinical history certifying that the deafness, arising at prelingual age, had prevented speech from developing; mean sensorineural hearing impairment ≥ 60 dB for 500-, 1,000- and 2,000-Hz frequency tones in the better ear. The information available for these subjects concerned: sex, date of birth and place of residence.

The number of residents per Region was calculated according to sex, age and residence, based on the latest national census performed in 2001 and updated to in 2003 [19].

The prevalence rates were assessed according to age and region. For areas showing a high prevalence, the rate assessment was more detailed and the local level was also considered. The homogeneous distribution, according to region and age bracket, was tested by chi-square test. The statistical analysis was performed using the SPSS statistics software [20].

Results

The results for 2003 showed 40,887 cases (20,915 males and 19,778 females) of prelingual sensorineural hearing impairment ≥ 60 dB, with a prevalence in the population of 0.72 per 1,000 inhabitants (Italian population = 56,780,305) [19]. The prevalence of deafness differs according to sex: 0.78 per 1,000 for males and 0.69 per 1,000 for females ($p < 0.001$).

As far as concerns geographic distribution, the prevalence rates of prelingual deafness showed significant differences ($p < 0.001$): 15,644 cases in North Italy (0.63 per 1,000), 7,111 in Central Italy (0.64 per 1,000), and 18,132 in South Italy and Islands (0.87 per 1,000) (Table I).

Deafness was found to be more frequently observed in the older age brackets, 50-64 years and > 64, living in Southern Italy and the islands, where the 1/1000 limit is exceeded by 1.27 and 1.15, respectively. For the 0-14 years age bracket, results concerning northern and southern regions

Table I. Geographic distribution of cases in Italy.

Age bracket (yrs)	Centre	North	South and islands	Total
0-14	418	1,130	1,2	2,820
15-49	3,386	7,485	8,702	19,573
50-64	1,799	3,602	4,367	9,768
≥ 64	1,508	3,427	3,791	8,726
Total	7,111	15,644	18,132	40,887

Table II. Prevalence rates of prelingual deafness in Italy according to age bracket and geographic area.

Age bracket (yrs)	North	Centre	South and islands	Total
0-14	0.37	0.29	0.3	0.35
15-49	0.63	0.63	0.83	0.71
50-64	0.73	0.83	1.27	0.93
≥ 64	0.70	0.68	1.15	0.84
Total	0.63	0.64	0.87	0.72

Regions
Rates of prevalence per 1,000

Piemonte	0.61
Valle d'Aosta	N.R.
Lombardia	0.62
Liguria	0.61
Trentino-Alto Adige	N.R.
Veneto	0.56
Friuli-Venezia Giulia	0.68
Emilia-Romagna	0.60
Toscana	0.61
Umbria	0.64
Marche	0.66
Lazio	0.71
Abruzzo	0.84
Molise	0.94
Campania	0.71
Puglia	0.82
Basilicata	1.17
Calabria	1.08
Sicilia	1.25
Sardegna	0.67

N.B. Valle d'Aosta and Trentino Alto Adige regions were not assessed for prevalence rate (W.N.).

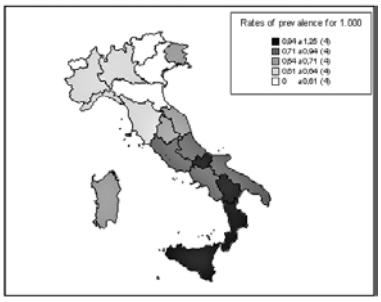

Fig. 2. Prevalence rate and distribution of prelingual deafness in the different Italian regions.

were homogeneous, whereas data related to central regions are significantly lower ($p < 0.001$) (Table II).
The results of prevalence, analysed for the separate regions, show a significantly heterogeneous distribution ($p < 0.001$). Basilicata, Calabria and Sicily show a higher prevalence of profound prelingual deafness; in those regions, the 1/1000 limit is exceeded by 1.17, 1.08 and 1.25, respectively (Fig. 2).
The prevalence of congenital deafness has been decreasing over the years; the 1.01/1000 ratio observed in the years 1934-1943 dropped to 0.55 in 1974-1988 and to 0.35 in 1989-2003 (Fig. 3).
When town data, related to these high-prevalence regions, were studied in depth, the rate referring to some rural areas was found to be 7 to 10 times higher than the national average (Fig. 4).

Conclusions

Very few scientific studies have been carried out in Italy on the prevalence of prelingual deafness. The results emerging from the present investigation reveal a national rate of prelingual hypoacusis of 0.7/1000.
The gradual and steady decrease observed, in recent decades, in the prevalence rates of the disease may result from a better prevention of infectious diseases in pregnancy and health surveillance for women of child-bearing age. At the same time, a decreased prevalence has been observed as

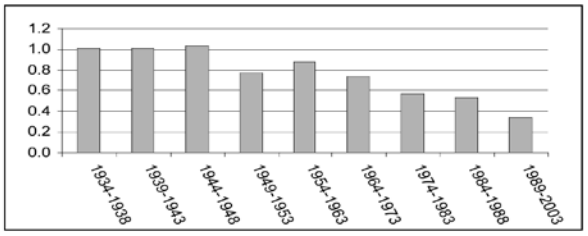

Fig. 3. Rates of prevalence per 1000, according to birth cohort.

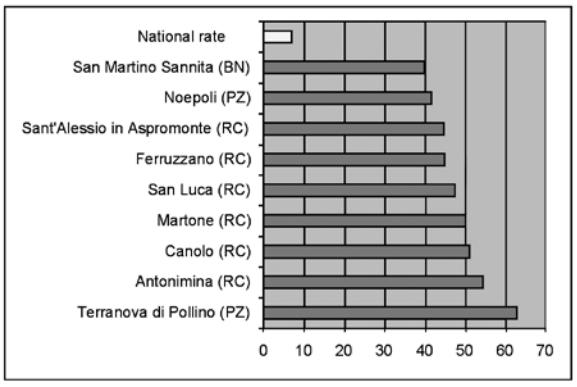

Fig. 4. Deafness prevalence rates in some Italian towns.

far as concerns rubeola, known to be a major aetiological cause of congenital deafness, following the introduction of rubeola vaccination, first recommended, in Italy, in 1972 [21].

The statistically significant sex-related differences may be partially explained by genetic factors, such as X-linked defects.

Analysis of results shows a clear heterogeneity in the prevalence rates involving the 50-64 and > 64-year-old age brackets living in the southern regions of Italy.

The prelingual hearing impairment was widespread in South Italy, particularly in some southern areas and islands where socio-economic development was lower. The 1/1000 prevalence rate, indicated by the WHO (World Health Organization), is especially exceeded in Basilicata, Calabria and Sicily [2].

Bearing in mind that, as observed by some Authors [22-24], the rate of profound deafness is higher in population groups in which consanguineous marriage is not uncommon and where lower socioeconomic development has taken place, the phenomenon can be easily explained.

In a recent study on consanguineous marriages, in Italy [25], data from the Vatican Archives on 520,492 consanguineous marriages, for which dispensation had been requested in the years 1911-1964, Cavalli Sforza et al. demonstrated that, in those same regions of Southern Italy (Basilicata, Calabria, Sicily), marriages with blood-related partners amounted to over 40% in the years 1935-1939.

Some factors, such as altitude, village size, population density, and migration, have a marked influence on the overall frequency of consanguineous marriages. When villages are small, the choice of a spouse, i.e., an unmarried person, who is of approximately the same age and also has other desirable requisites may be limited, within the village to very few individuals, possibly between zero and five [26 27].

Increased migratory movements, the changes in communication, transportation, work availability, improved education and, indeed, also the social contacts which occurred in the Twentieth Century resulted in a rapid decrease in consanguineous marriages. Likewise, the prevalence of congenital deafness has also decreased; the 1.01/1000 ratio observed in the years 1934-1943 had dropped to 0.55 in 1974-1988 and to 0.35 in 1989-2003 (Fig. 3).

For the 0-14-year-old age bracket, in fact, results from northern and southern regions are more homogeneous (Table II).

It is worthwhile stressing that in our study, the rate observed in the general population refers only to cases of bilateral prelingual deafness ≥ 60 dB established using rigorous diagnostic criteria. In particular, the validation criteria of hearing disability involving communication handicaps, from 1992 to 2003, was restricted by the Ministry of Economy, which checked the authenticity of disability through a Supreme Medical Committee [17 18].

The prevalence reported may, therefore, appear to be lower than that obtained in other epidemiological studies carried out on restricted population samples and with a different approach to assessing the deafness level which not necessarily results in a communication handicap.

As no previous population data are available, the results reported may prove helpful in monitoring the disease trend and in predisposing effective and targeted national and regional prevention plans. No doubt, the high rate identified in certain small rural Communities in the South of Italy needs to be further studied in depth in order to corroborate the existence of possible genetic clusters.

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