Original article

AUTISM SPECTRUM DISORDER IN ITALY: DEMAND FOR AN INTEGRATED EPIDEMIOLOGICAL SURVEILLANCE SYSTEM.

Walter Mazzucco[1], Michele Tumminello[2], Rosaria Nardello[1], Ornella Giambalvo[2], Vincenzo Cicala[3], Lavinia Lo Bianco[3], Antonietta Turrisi[4], Alessandra Casuccio[1], Gianna Agrò[2] AND the Multidisciplinary and Epidemiological Studies on Autism spectrum disorder (MESA) Group.

Summary

Autism spectrum disorder (ASD) is a complex neurodevelopmental syndrome of emerging public health concern, according to a documented significant increase of diagnosed cases of ASD in Europe and USA.

In Italy, actually, it is not possible to estimate at national level a reliable ASD occurrence by using existing health and scholastic data flows. The lack of information has implications on social and healthcare services dedicated to subjects affected by ADS.

The database of the Italian institute in charge of social and security assistance was accessed at the provincial level to investigate the ASD cases occurred in the Palermo province. The official reports of all subjects visited in 2013 by INPS physicians were analyzed by using an automatic software and diagnosis consistent with ASD were extracted and flagged.

Our findings support the choice of alternative use of INPS administrative database in order to define a reliable ASD occurrence estimate as first step to develop an integrated epidemiological surveillance system on ASD.

Background

Autism spectrum disorder (ASD) is a complex neurodevelopmental syndrome characterized by impairments in two major domains: deficits in social communication and reciprocal social interaction and restricted repetitive and stereotyped patterns of behaviour, interests and activities [1]. A number of rare gene changes or mutations have been identified in association with ASD, but there are several evidences in sup-

Address of the authors:
1. *Dipartimento Scienze per la Promozione della Salute e Materno Infantile (PROSAMI) "G. D'Alessandro" - University of Palermo, Palermo, Italy.*
2. *Dipartimento Scienze Economiche, Aziendali e Statistiche (SEAS) – University of Palermo, Palermo, Italy.*
3. *Specialization School in Infant Neuropsychiatry, University of Palermo Medical School, Palermo, Italy.*
4. *Centro Medico Legale Istituto Nazionale Previdenza Sociale (INPS) Palermo, Palermo, Italy.*

Send correspondence to: *Walter Mazzucco, walter.mazzucco@unipa.it*

port of potential environmental risk factors (pollutants, diet, etc.) influencing early brain developmenton in combination to a polygenic basis. [2,3]

ASD global estimated prevalence among live-born children is 1% [4]. Recent population-based studies have documented in some areas of Asia, Europe and North America, ASD prevalence estimates of over two percent among live-born children [5,6]. Centers for Diseases Control and Prevention (CDC) have estimated in the United States an ASD occurrence of ASD cases of 4.5 times higher among boys than among girls [7].

In the last decades, epidemiological data have shown a significant increase of diagnosed cases of ASD both in Europe and USA, accounting for a raise in prevalence and incidence with almost an equivalent decrease in diagnoses of mental retardation cases [8-13], partially due to a better definition of the diagnostic criteria and a greater focus on neurodevelopmental disorders [14-17].

The raising of ASD observed cases has required a rapid and deep process of healthcare services reorganization, both in terms of an anticipation of the age of diagnosis through standardized diagnostic criteria and of services access and continuity between diagnosis and the beginning of an adequate integrated therapeutic plan. Such a multidisciplinary assessment of the health demand and of healthcare service is essential in order to make early interventions addressed to both the behavioural and social sphere, which have, in turn, proven effectiveness in reducing symptoms of anxiety and aggression, improving quality of life of individuals and their families, and promoting the social inclusion of people affected by ASD [18].

In Italy, ASD epidemiological data have been provided on regional level and only by few Regions, documenting prevalence estimates slightly under Europe and USA [19]. Anyway, actually, it is not possible to estimate reliable data at national level on ASD occurrence and impact by using existing health and scholastic information systems. The lack of information is due both to limitations in the integration of different data flows and to the evidence that ASD patients are mainly assisted in outpatient settings. Also, the delay in the diagnosis of ASD patients can result in misdiagnosis according to the different clinical presentation of ASD in adolescents or adult patients.

Thus, the lack of an integrated information system could in part explain the under development in Italy of services, programs and integrated social and healthcare pathways, dedicated to the support of ASD patients and their families, from early diagnosis to rehabilitation and social integration. Another documented critical point is represented by the transition from adolescence to adult age in ensuring both an healthcare continuity and a global approach to the person with ASD.

The higher perception of ASD increasing over time suggests the opportunity to define a dedicated information system in order to estimate its epidemiological impact in Italy.

Aim of this study is to explore at provincial level an alternative use of an existing administrative font, the database of the Italian institute in charge of social and security assistance, in order to define a reliable ASD occurrence estimate as a first step to develop an integrated epidemiological surveillance system on ASD.

Material and methods

A multidisciplinary research group, named Multidisciplinary and Epidemiological Studies on Autism spectrum disorder (MESA) Group, composed by public health and infant neuropsychiatric physicians, statisticians, mathematics, geneticists and economists, was created within University of Palermo.

The "Istituto Nszionale per la Previdenza Sociale" (INPS) is a public institution in charge of social security and assistance services dedicated to employees, public or private, and autonomous workers or to citizens affected by disabilities [20]. Together with the National Health System (NHS), INPS represents the Italian welfare state [21].

INPS social assistance services consist in the provision of financial support, which is differentiated, according to the degree of disability or impairment and to specific requirements, in partial and total

attendance allowance. Partial attendance allowance is attributed on demand to low income citizens aged under 18 and affected by disability (subjects with persistent difficulties in carrying out tasks and functions typical of their age) [22]. Total attendance allowance is a full financial support recognised to all not self-sufficient citizens affected by permanent disability (subjects with persistent limitations in carrying out normal and daily tasks and functions) [23]. Health status and disability degree is defined on the basis of a first medical examination performed by an integrated medical commission (IMC), composed by physicians designed by the local health authority (ASL) and by a forensic physician from the INPS district Office. Health reports are then validated by the INPS Legal Medical Center (CML). As medical disabilities may be susceptible to changes over time, assisted individuals are subjects to annual programmed or to not programmed sample medical revision examinations to confirm or revoke the financial support [24].

After a preliminary study on the available literature on ASD occurrence and epidemiology, the study group signed a formal scientific protocol with the Palermo district INPS Office in order to access the official reports of any potential ASD subject accessing to financial benefit, diagnosed by the first medical examination or by a medical revision examinations during 2013, to retrospectively estimate the ASD cases distribution of ASD cases occurrence in the Palermo city and in all the remaining 81 municipalities of the Palermo province. Each municipality was identified through the different postal codes.

As the INPS database was conceived for administrative purposes, it can be explored only by using the individual social security code or name. For this reason, the INPS database was queried by an *ad hoc* created procedure, described as follows: 1) selection of all the visited individuals by birth year (between 1995 and 2013, according to pediatric age extension), identified by the social security code; 2) development of a dedicated an algorithm by using Mathematica software

[25], to automatically open, read and extract the official reports available in .pdf format of the visited subjects; a selection of specific keywords (autism, disorder, cognitive, impairment, psychological, development, stereotype, alteration), single and in combination, were used; 3) access to the reports to identify the ASD individuals and the non ASD ones. Discordant diagnosis have been solved by a further access to the official reports in charge of specialized physicians.

Finally, for each one of the selected subjects, the following demographic variables were extracted for the study purposes: sex, age, birthplace and residence.

Descriptive statistics and graphics were performed by using Mathematica software [25].

Results

During 2013 the INPS database recorded official reports regarding 2931 individuals under 18 years old with a confirmed access to financial benefit. Individuals with an ASD diagnosis were 297 (10,1%). Sex ratio was about 4 to 1 (males= n. 234; 78,8%), result in the with literature [7]. Subjects with a total attendance allowance, because of diseases at greater severity [23], were 531 (18,1%). Of these, 165 (31%) were individuals documenting a diagnosis of ASD.

The distribution by year of birth, sex and diagnosis (ASD cases versus non ASD cases) of the 2931 individuals accessing to partial and total attendance allowance is presented in Figure 1. The distribution per year of ASD cases is greater in the period 2005-2010 as compared to 1999-2004 period, while non ASD cases tend to a decrease over time. As ASDs symptoms usually occur after 18 months of age, with an average age of diagnosis between 24 and 30 months, data on benefit concessions from 2011 to 2013 are not consistent as expected.

Figure 2 represents the distribution of all the visited individuals by diagnosis (ASD cases versus non ASD cases) and residence defined by postal code (Palermo city: from 90121 to 90151; other municipalities of the province: from 90010 to 90049). The distribution of the visited

individuals, with and without ASD, is not uniform among the municipalities of the

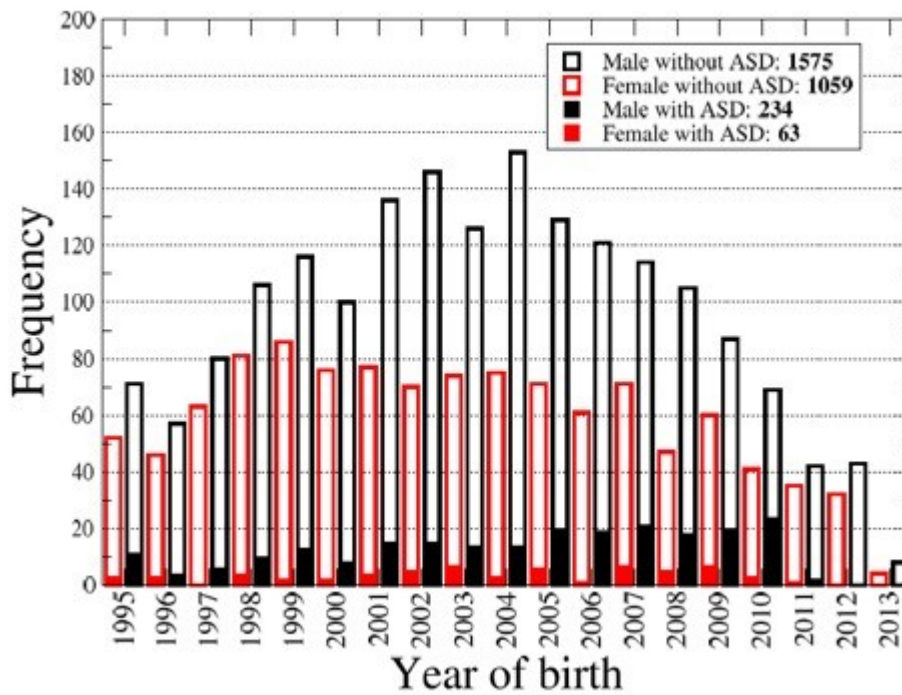province (left side of the figure) as well as in the different areas of the Palermo

Figure 1. Distribution of the 2931 visited individuals (partial and total attendance allowance) by year of birth, gender and diagnosis (ASD versus non ASD).
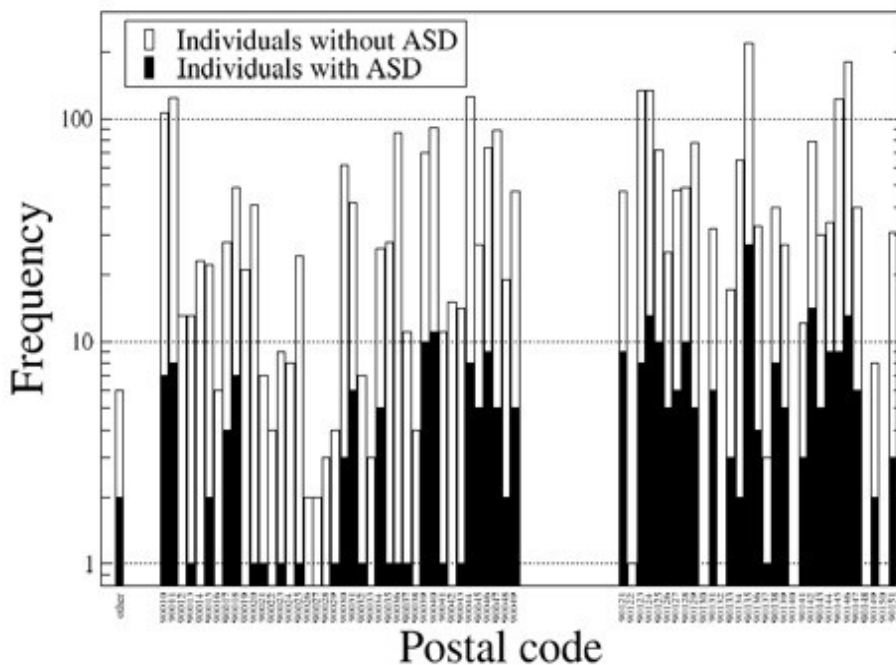

Figure 2. Distribution of the 2931 visited individuals (partial and total attendance allowance) by diagnosis (ASD versus non ASD) and residence defined by postal code (Palermo city: from 90121 to 90151 - right side of the figure; other municipalities of the province: 90010 to 90049 – left side of the figure).

city (right side of the figure), though it is noted that the areas most impacted are typically adjacent.

Figure 3 shows the distribution of the 531 individuals with total attendance allowance only by sex and place of residence (Palermo city versus Palermo province municipalities). The proportion of females diagnosed with ASD residing in the Palermo city (25%) is twice than the province (12%).

Discussion

This case report study has highlighted the importance to define an integrated epidemiological surveillance system on ASD in Italy.

According to the current fragmentary epidemiological picture on ASD in Italy, we have explored at a district level an alternative use of the Italian institute in charge of social and security assistance database, an existing administrative font used to collect data on social security and assistance services provided by the welfare system.

We documented an increase in the distribution per year of ASD cases. Moreover, descriptive results suggested to further investigate for association between ASDs occurrence and potential environmental variables [3] with regard to different expositions related to urban and not urban residence.

Our findings support the choice of alternative use of INPS administrative database in order to define a reliable ASD occurrence estimate as a first step to develop an integrated epidemiological surveillance system on ASD.

A systematic review of prevalence studies conducted by Williams JG et al. in 2006 has contributed to identifying some of the factors affecting the prevalence estimates. Particularly, over half of thevariation among study estimates could be explained by the age of the children screened, the diagnosticcriteria used, and the country studied, as well as the rural or urban location and whether cases were
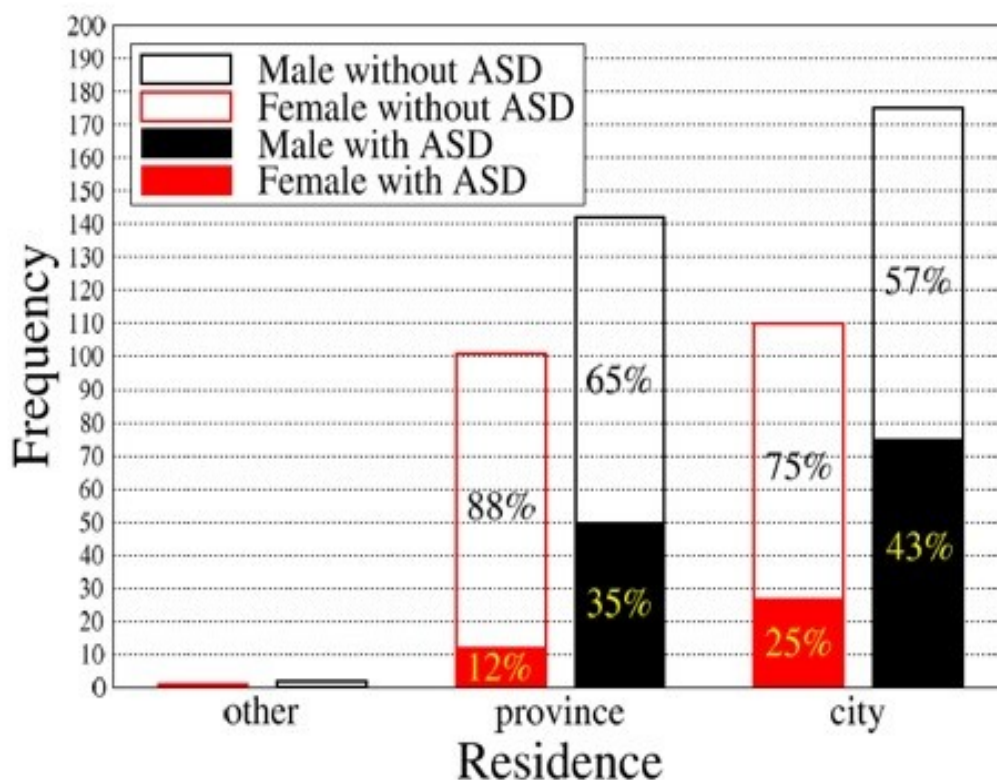

Figure 3. Distribution of the 531 visited individuals with total attendance allowance only by gender, residence (Palermo city versus remaining Palermo province municipalities) and diagnosis (ASD versus non ASD).

assessed prospectively or retrospectively. Consequently, a need to further investigate the impact of theseknown factors on prevalence estimates has been clearly documented [26].

In the same direction, in 2005 the Health and Consumer Protection Directorate – General of the European Commission stated the need of a more complex monitoring systems on AS [27].

Thus, future steps will consist in the integration of the obtained estimates with data provided by existing health and scholastic information systems, to better define, for instance, the proportion between ASD, Asperger's syndrome and pervasive developmental disorder-not otherwise specified (PDD-NOS) cases [9].

Therefore, the implementation of an integrated epidemiological surveillance system on ASD has to be considered strategic in order to develop an appropriate clinical governance based on multidisciplinary networks and to facilitate the coordination of regional public health institutions and privates, overcoming any discrimination based on the access to social and healthcare services. This approach will also support the definition of specific guidelines for early ASD diagnosis, treatment and rehabilitation, as well as specific pathways and dedicated services. Lastly, improving the integration of the existing data flows will promote basic, clinical and epidemiological research in order to increase the general level of knowledge of ASD disorders [28].

As ASDs represent an emergent public health priority, all the mentioned expectations are to be considered in line with recent legislative action taken both by the European Parliament [29] and by the Italian Parliament [30].

Although the present study presents several limitation related to the administrative use of the INPS databases, the alternative use of the INPS social security and assistance data flow can be considered a starting point to perform prevalence study and to integrate the different existing health and scholastic information systems. Future study will be performed to explore the application of the proposed model both at regional and national level.

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