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A screening on Specific Learning Disorders in an Italian speaking high genetic homogeneity area

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ABSTRACT

The aim of the present research is to investigate the prevalence of Specific Learning Disorders (SLD) in Ogliastra, an area of the island of Sardinia, Italy. Having experienced centuries of isolation, Ogliastra has become a high genetic homogeneity area, and is considered particularly interesting for studies on different kinds of pathologies.

Here we are going to describe the results of a screening carried out throughout 2 consecutive years in 49 second grade classes (24 considered in the first year and 25 in the second year of the study) of the Ogliastra region. A total of 610 pupils (average age 7.54 years; 293 female, 317 male) corresponding to 68.69% of all pupils who were attending second grade in the area, took part in the study.

The tool used for the screening was "RSR-DSA. Questionnaire for the detection of learning difficulties and disorders", which allowed the identification of 83 subjects at risk (13.61% of the whole sample involved in the study). These subjects took part in an enhancement training program of about 6 months. After the program, pupils underwent assessment for reading, writing and calculation abilities, as well as cognitive assessment.

According to the results of the assessment, the prevalence of SLDs is 6.06%. For what concerns dyslexia, 4.75% of the total sample manifested this disorder either in isolation or in comorbidity with other disorders. According to the first national epidemiological investigation carried out in Italy, the prevalence of dyslexia is 3.1–3.2%, which is lower than the prevalence obtained in the present study.

Given the genetic basis of SLDs, this result, together with the presence of several cases of SLD in isolation (17.14%) and with a 3:1 ratio of males to females diagnosed with a SLD, was to be expected in a sample coming from a high genetic homogeneity area.

1. Introduction

Specific Learning Disorders (SLD) are defined as disorders that have a neurobiological origin and that are independent from environmental, socio-cultural, or psychological factors. They are "specific" in the sense that they affect specific ability

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domains in a significant but circumscribed way, while leaving intact the individual's cognitive functioning. Specific Learning Disorders are also independent from any sensorial deficits and are developmental in nature. This means that they become evident during developmental age, when the child is exposed to literacy learning, but also that the developmental character of these disorders is related to the continuously changing and evolving neural structures of the child.

Specific Learning Disorders include dyslexia, dysgraphia, dysortography and dyscalculia. According to the International Classification of Diseases (ICD-10) of the World Health Organization, currently adopted in Italy, dyslexia, dysortography and dyscalculia are defined respectively as specific reading disorder (F81.0), specific spelling disorder (F81.1) and mathematics disorder (F81.2); dysgraphia, a writing disorder that concerns the fine motor skills of handwriting, is included among other developmental disorders of scholastic skills (F81.8). SLDs can be associated with each other and/or with other developmental disorders like dyspraxia, attention deficit disorders, language disorder, etc. They are very heterogeneous, both in terms of manifestations and of functional individual profiles.

A variety of hypotheses can be found in the literature about the origin of SLDs. Research on dyslexia, the SLD that has been investigated for longer than others, has identified several factors that can be considered possible causes: phonological disorders (Ramus, 2003; Snowling, 2001), the lack of automation of the connection between visual perception and language (Tallal, Miller, & Fitch, 1993), specific visuoperceptual disorders and magnocellular system disorders (Best & Demb, 1999; Livingstone, Rosen, Drislane, & Galaburda, 1991; Stein, 2001). The traditional distinction between "core deficit" symptoms and secondary symptoms has been criticised (Nicolson & Fawcett, 2007) and, to date, wide agreement has been reached on the idea of a multifactorial origin of SLDs (Bishop, 2015; Gooch, Hulme, Nash, & Snowling, 2013; Pennington, 2006; van Bergen, van der Leij, & De Jong, 2014). The appropriateness of the term "specific disorder" in itself is beginning to be questioned, especially in the light of the great comorbidity that can be observed both within SLDs (homotypic comorbidity) and between SLDs and other developmental disorders, like ADHD, DCD, dyspraxia (heterotypic comorbidity). Pennington (2006) has provided the greatest contribution to the idea of a multifactorial origin of SLDs and, starting from his model, Wolf (2007), has elaborated the so-called "pyramid" of the behaviours of reading. This can be seen as a sort of iceberg, in which the upper, visible part corresponds to the observable difficulties manifested by the child. These are subject to influence from the "environment" (education received, socio-cultural context, etc.) and can be present despite good general cognitive abilities. The manifested difficulties can concern reading or other basic learning skills. The invisible part of the "pyramid" contains the underlying cognitive processes (perceptual, linguistic, motor, mnemonic, attentive) and executive functions, that determine the performance examined and assessed at a neuropsychological level. The variety and complexity of these processes, and of their interactions, can explain how the same "symptom" (deficit in reading or writing) may be determined by a variety of factors and by the different interactions among them. Further down, the "pyramid" contains the structures of the neural networks, made up of the neurons and of their synaptic circuits. Finally, the base of the "pyramid" contains the genetic basis, responsible for the development of the cells of our body, in interaction with the particular life environment.

Recent research studies have shown that reading does not have a Mendelian hereditary basis and that there is no single specific "gene for dyslexia" (Bishop, 2015). Studies on the familiarity in dyslexia have identified multiple genetic loci (Becker et al., 2014; Grigorenko, 2005; van Bergen et al., 2014). The variety of genetic loci is what determines the variability of the neuropsychological deficits that can be observed, and consequently the subtypes of this SLD that may manifest themselves as different phenotypes. For a wide and comprehensive review on the relation between dyslexia and genetics, and on the interplay between genetics and the environment in determining reading skills see Peterson and Pennington (2015).

As a matter of facts, researchers have now reached an agreement on the fact that besides the neurobiological variability of SLDs, the environmental variability also needs to be considered (see Bishop, 2015). On the one hand, dyslexia and other SLDs have a genetic origin (see, among others Becker et al., 2014; Paracchini, Scerri, & Monaco, 2007; Shalev et al., 2001), in this sense being independent from the environment, on the other hand, the environment can significantly influence the way and the extent to which the dyslexic pupil can compensate for his/her disorder. Children with a SLD need to "adapt" to the environment, and this is where school and school professionals can intervene to favour and enhance learning. To this purpose, it is important to carry out an early screening in order to promptly activate support measures.

In this perspective, the aim of the present study is twofold: it intends to carry out an early screening on Specific Learning Disorders (dyslexia, dysortography, dysgraphia and dyscalculia) while investigating their prevalence in a high genetic homogeneity area. The latter is Ogliastra, a region in the central-east part of the Sardinia island, Italy. Fig. 1 represents the investigation area and contains the districts that took part in the study. The districts are 23 in total, and in some cases they include more than one village.

For centuries, Ogliastra has experienced isolation, it has been subject to little influence from outside and has undergone little change. As a consequence, the population of this area is among those with highest genetic homogeneity in Europe (Cavalli-Sforza, Menozzi, & Piazza, 1994; Jorde, Aw, Morgan, & Workman, 1982; Piazza, 1993), together with the Icelandic, the Sami and the Basque populations. Since these people have remained geographically and/or culturally and/or demographically isolated for centuries, endogamy has limited the set of genes transmitted from a generation to another. This has favoured high levels of homozygosity and the emergence of recessive characteristics.

People living in Ogliastra represent one of the most closest communities of Sardinia (Pistis et al., 2009). Immigration phenomena here have started to take place only very recently and the community is considered the most conservative of the island (Zei et al., 2003). Ogliastra is also the area with the lowest population density in Italy (about 31.26 people per km^2) and, as already mentioned, with a very high percentage of endogamy. For example, in one of the villages of the area, Talana, about 80% of residents belong to only 8 paternal and 11 maternal lineages (Angius et al., 2001).