A methodology for the characterization and diagnosis of cognitive impairments—Application to specific language impairment

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ABSTRACT

Objectives: The diagnosis of mental disorders is in most cases very difficult because of the high heterogeneity and overlap between associated cognitive impairments. Furthermore, early and individualized diagnosis is crucial. In this paper, we propose a methodology to support the individualized characterization and diagnosis of cognitive impairments. The methodology can also be used as a test platform for existing theories on the causes of the impairments. We use computational cognitive modeling to gather information on the cognitive mechanisms underlying normal and impaired behavior. We then use this information to feed machine-learning algorithms to individually characterize the impairment and to differentiate between normal and impaired behavior. We apply the methodology to the particular case of specific language impairment (SLI) in Spanish-speaking children.

Methods and materials: The proposed methodology begins by defining a task in which normal and individuals with impairment present behavioral differences. Next we build a computational cognitive model of that task and individualize it: we build a cognitive model for each participant and optimize its parameter values to fit the behavior of each participant. Finally, we use the optimized parameter values to feed different machine learning algorithms. The methodology was applied to an existing database of 48 Spanish-speaking children (24 normal and 24 SLI children) using clustering techniques for the characterization, and different classifier techniques for the diagnosis.

Results: The characterization results show three well-differentiated groups that can be associated with the three main theories on SLI. Using a leave-one-subject-out testing methodology, all the classifiers except the DT produced sensitivity, specificity and area under curve values above 90%, reaching 100% in some cases.

Conclusions: The results show that our methodology is able to find relevant information on the underlying cognitive mechanisms and to use it appropriately to provide better diagnosis than existing techniques. It is also worth noting that the individualized characterization obtained using our methodology could be extremely helpful in designing individualized therapies. Moreover, the proposed methodology could be easily extended to other languages and even to other cognitive impairments not necessarily related to language.

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1. Introduction

The characterization and diagnosis of cognitive impairments are, in most cases, problematic. There are two main reasons for their difficulties: heterogeneity and overlap. Many cognitive impairments present a general behavioral profile inside which we can find many different behavioral trends for each patient. These individual differences are crucial for the design of personalized therapies and could provide important clues to better understand each deficit. Clinical evidence shows the effectiveness of early and individualized intervention for very different cognitive impairments and, in particular, for many communication disorders [1]. However, there are very few studies that consider these individual differences. The other main problem is the high overlap presented by many cognitive impairments. Many disorders co-occur and share similar symptoms and some aspects of their behavioral profiles.

All these factors motivate the design and development of new methodologies for the characterization and diagnosis of cognitive impairments that consider individual differences. Existing techniques just focus on behavioral variables. However, as stated above, behavior is in most cases heterogeneous and is shared by many

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cognitive impairments. Therefore, focusing solely on behavioral variables might not be the most appropriate way to design such methodologies. It might be more appropriate to investigate the cognitive mechanisms underlying normal and impaired behavior. Finding patterns at the cognitive level might be very useful for the characterization and diagnosis of the impairment. The tool we propose to investigate the underlying cognitive mechanisms is computational cognitive modeling.

In this paper, we propose a methodology for the characterization and diagnosis of cognitive impairments and apply it to the particular case of specific language impairment (SLI) in Spanish-speaking children. The approach is based on building a computational cognitive model of a task in which children with impairment show differences with normal children. Then the different parameters of the model are used to train machine-learning algorithms to support the processes of diagnosis and characterization. We applied the methodology to an existing database of Spanish-speaking children. The results show that the information obtained from the computational cognitive models is highly informative for characterization and diagnosis. Additionally, the results show the utility of this methodology to support or reject existing theories on the causes of the impairment. It is also worth mentioning that this methodology could easily be extended to other languages and even to other cognitive impairments not necessarily related to language.

2. Background

SLI is generally defined as a developmental disorder of language ability in the absence of factors that usually affect language learning, such as hearing impairments, low non-verbal intelligence or neurological damage [2]. Numerous studies have investigated the cognitive profile of children with SLI and have attempted to characterize the impairment and establish a standardized diagnosis mechanism. However, none of these objectives have been achieved to date. In the following subsections we briefly review the existing theories on the disorder and the common diagnostic techniques.

2.1. Existent theories on SLI

SLI has been extensively studied from many different perspectives. However, there is no unified account for its particular developmental profile. There are two main reasons for this lack: heterogeneity and overlap. SLI presents a broadly heterogeneous profile [2]. Different individuals tend to show wide differences in the severity of the disorder, the areas of language affected by the disorder and how these areas are affected within the same child over time [3]. This variation has led to the definition of different SLI subgroups and even different definitions for each individual profile within SLI [4,5]. The other main reason is the high overlap of SLI with other cognitive impairments. SLI co-occurs with certain other disorders such as semantic-pragmatic disorder and the autistic spectrum [4], attention deficit hyperactivity disorder [6], and some motor-related disorders [7]. Furthermore, contrary the first theories that tried to account for SLI, it has now been proved that this impairment is not just restricted to language. Many other cognitive functions are impaired such as working memory [8] and some motor skills [7]. This high heterogeneity and overlap make it very difficult to differentiate SLI from other cognitive impairments and to distinguish between different subcategories grouped under the SLI profile. Therefore, there is no unified account for the particular developmental profile of SLI. There exist three main theories that attempt to account for this developmental profile: the grammar specific deficit (GSD), the processing deficit (PD) and the procedural deficit hypothesis (PDH).

2.1.1. Grammar specific deficit

GSD theories consider SLI to be due to a deficit in the processing of grammar. The most famous example of this theory is most likely the extended optional infinitive (EOI) hypothesis [9]. Typically developing children go through a stage in which they omit verbal suffixes such as tense, person and number agreement markers [10]. The EOI hypothesis argues that this stage of immature grammar is much longer in SLI children. Other authors claim that the problems with grammar are due to the under-representation of grammatical and linguistic features [11,5]. Although these theories account quite well for the language profile of SLI children, they cannot explain all the linguistic phenomena detected either in English or in other languages. Moreover, any purely grammatical explanation cannot account for the problems presented by SLI children when performing non-linguistic tasks.

2.1.2. Processing deficit

Some authors attribute the problems associated with SLI to a processing deficit not related to language. This particular processing deficit could be general or specific to very concrete systems such as the phonological system (see [12] for a brief review). General processing deficit theories attribute the problems of SLI children to a somewhat limited processing capacity [13,14]. The generality of the impairment proposed by these theories explains many of the deficits of the SLI profile. However, they fail to account for some linguistic and non-linguistic impairments [2,15]. There are also some processing deficit theories that consider that the SLI profile to be caused by a specific deficit localized in the phonological system, affecting the working memory [16]. Again, these hypotheses fail to account for all the impaired linguistic and non-linguistic functions found in SLI [2,15].

2.1.3. Procedural deficit hypothesis

According to the declarative-procedural model [17], there exists a dissociation in the cognitive system between declarative and procedural memory. Declarative memory stores semantic knowledge, while procedural memory handles learning, grammar and rule-based skills. The PDH [18] is an extension of this theory that proposes that SLI is basically a disorder of the procedural system. Declarative memory tries to compensate for this deficit, but the result is still an impaired developmental profile [15]. Thus, this model explains the linguistic problems related to the acquisition of syntax and also the motor deficits found in SLI, (by maintaining that those deficits are related to implicit sequence learning tasks that rely on the procedural system). The PDH seems to be the most complete account of SLI to date. However, some authors note that not all of its conclusions are necessarily true [19] and that a statistical learning account of SLI could better explain the behavioral data [20].

All the existing theories capture some features of the behavioral profile of SLI. However, there is no hypothesis that accounts for the wide range of linguistic and non-linguistic problems found in SLI and for the individual differences present in the SLI profile. This fact could suggest the existence of different subtypes of SLI and the consequent need for individualized diagnostic techniques.

2.2. Current diagnostic techniques

SLI diagnosis is usually based on both inclusionary and exclusionary criteria. The general approach for assessing language development uses language tests such as inflection tasks, vocabulary tests or non-word repetition tasks, among many others [2]. Typically, children performing significantly lower than average on two or more of these measures are diagnosed as impaired. Some other observable variables are taken from transcriptions of children's spontaneous speech, such as the mean length of utterance